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2 BIOLOGICAL AND ENVIRONMENTAL FOUNDATIONS AND PRENATAL DEVELOPMENT

LEARNING OBJECTIVES

- **2.1** Discuss the genetic foundations of development.
- **2.2** Identify examples of genetic disorders and chromosomal abnormalities.
- **2.3** Describe behavior genetics and the ways in which heredity and environment interact to influence development.
- **2.4** Summarize three periods of prenatal development and commonly used prenatal diagnostic tests.
- **2.5** Analyze the influence of teratogens on prenatal development.
- **2.6** Examine the influence of parental characteristics and prenatal care on development.
- **2.7** Expain the process of childbirth.

"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

What determines our traits, such as appearance, physical characteristics, health, and personality? We are born with a hereditary "blueprint" that influences our development. As discussed in the following sections, this blueprint is inherited from our biological parents.

Genetics

The human body is composed of trillions of units called cells, each with a nucleus containing 23 matching pairs of rod-shaped structures called **chromosomes** (Finegold, 2019). 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. **Genes** carry the plan for creating all of the traits that organisms carry. An estimated 20,000 to 25,000 genes reside within the chromosomes, comprising the human genome and influencing all genetic characteristics (Taneri et al., 2020).

Much of our genetic material is not unique to humans. Every species has a different genome, yet we share some genes with all organisms, from bacteria to primates. We share nearly 99% of our DNA with our closest genetic relative, the chimpanzee. There is even less

genetic variation among humans. People around the world share 99.9% of their genes (Lewis, 2017; National Human Genome Research Institute, 2018). Although all humans share the same basic genome, every person has a slightly different code, making them 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, duplicating chromosomes, resulting in new cells with identical genetic material (Sadler, 2018). The process of mitosis accounts for the replication of all body cells.

Sex cells reproduce differently, through **meiosis**. First, the 46 chromosomes begin to replicate as in mitosis, duplicating themselves. But before the cell completes dividing, a critical process called *crossing over* takes place. The chromosome pairs align and DNA segments cross over, moving from one member of the pair to the other, essentially "mixing up" the DNA. Crossing over thereby creates unique combinations of genes (Finegold, 2019). The resulting cell consists of only 23 single, unpaired chromosomes. Known as **gametes**, these cells are specialized for sexual reproduction: sperm in males and ova in females. Ova and sperm join at fertilization to produce a fertilized egg, or **zygote**, with 46 chromosomes, forming 23 pairs with half from the biological mother and half from the biological father. Each gamete has a unique genetic profile and ndividuals can produce millions of genetically different gametes (U.S. National Library of Medicine, 2020).

Sex Determination

The sex chromosomes determine whether a zygote will develop into a male or female. Twenty-two of the 23 pairs of chromosomes are matched pairs (Figure 2.1). They contain similar genes in almost identical positions and sequence, reflecting the distinct genetic blueprint of the biological mother and father. The 23rd pair of chromosomes are not identical because they are sex chromosomes that specify the genetic 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).

Because females have two X sex chromosomes, all their ova contain one X chromosome. A male's sex chromosome pair includes both X and Y chromosomes; therefore, one half of the sperm males produce contain an X chromosome and one half contain a Y. The Y chromosome contains genetic instructions that will cause the fetus to develop male reproductive organs. Thus, 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 (Figure 2.2).

Genes Shared by Twins

All biological siblings share the same biological parents, inheriting chromosomes from each. Despite this genetic similarity, siblings are often quite different from one another. Twins are siblings who share the same womb. Twins occur in about 1 out of every 33 births in the United States (J. A. Martin et al., 2018).

Monozygotic, or identical, twins share 100% of their DNA.

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FIGURE 2.1 ■ Chromosomes

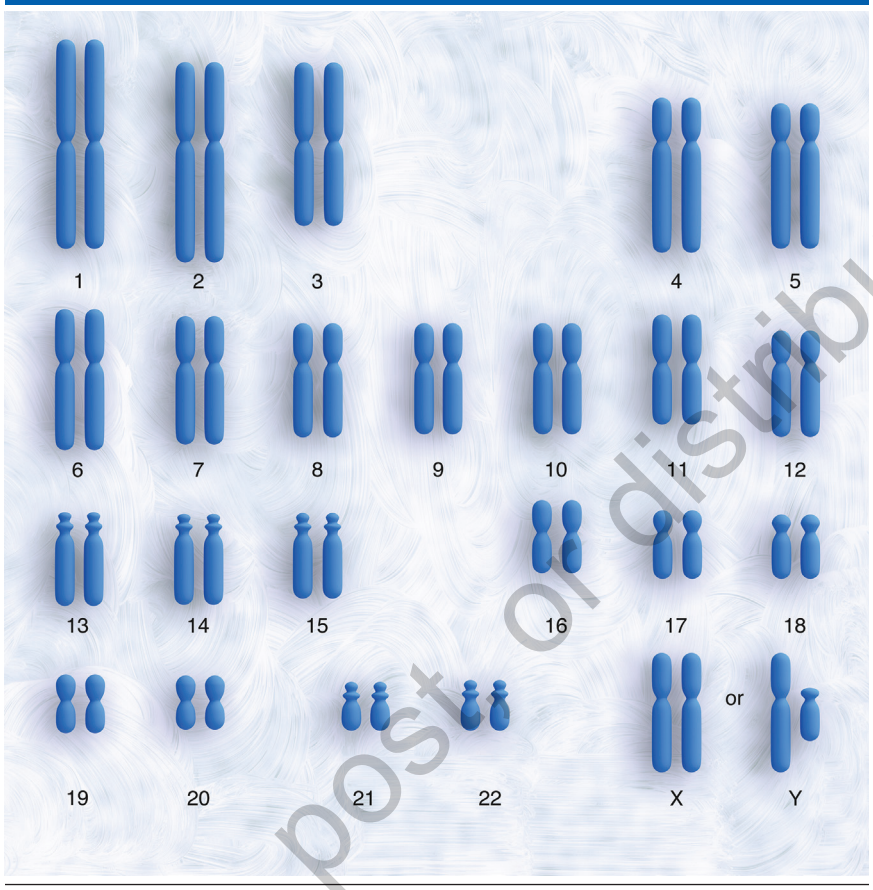

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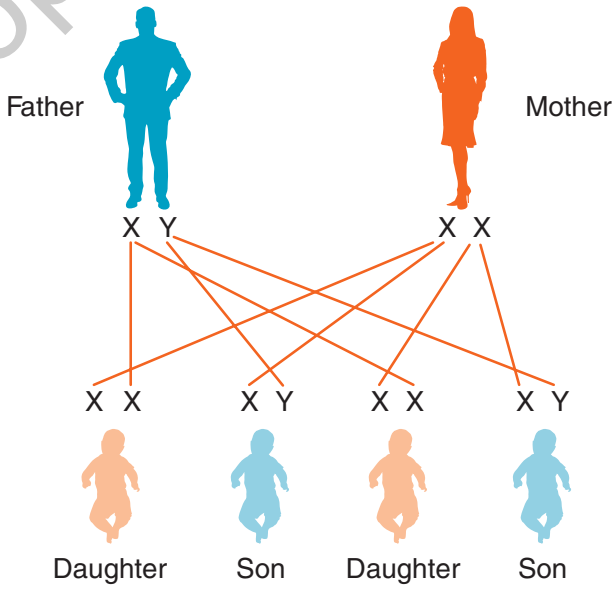
FIGURE 2.2 ■ Sex Determination



The majority of naturally conceived twins (over 70%) are **dizygotic (DZ) twins,** or fraternal twins, created when a woman releases more than one ovum and each is fertilized by a different sperm (Gill et al., 2019). DZ twins share about one half of their genes and, like other siblings, most fraternal twins differ in appearance, such as 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 (Hazel et al., 2020). Rates of DZ twins also increase with in vitro fertilization, maternal age, and with each subsequent birth (Gill et al., 2019; Pison et al., 2015).

Monozygotic (MZ) twins, or identical twins, originate from the same zygote, sharing the same **genotype**, or set of genetic instructions for all physical and psychological characteristics. MZ twins occur when the zygote splits into two distinct separate but identical zygotes that develop into two infants. MZ twins are estimated to occur in 1 in every 250 births (American College of Obstetricians and Gynecologists & Society for Maternal-Fetal Medicine, 2014). The causes of MZ twinning are not well understood (McNamara et al., 2016). Rates of MZ twins are not related to maternal age or the number of births, but MZ twins are more likely to occur with vitro fertilization (Busnelli et al., 2019; 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. Some 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? Some traits, like hair color, are passed through **dominant–recessive inheritance**. As we have discussed, each person has 23 pairs of chromosomes, half inherited from the biological mother and half from the biological father. Some genes are *dominant* and are always expressed regardless of the gene they are paired with. Nonred hair is a dominant gene. Other genes, such as for red hair, are *recessive* and are only expressed if paired with another recessive gene.

For example, suppose two biological parents each carried a dominant gene for nonred hair (symbolized by N in Figure 2.3) and a recessive gene for red hair (r). Since dominant genes override recessive genes, both biological parents will have nonred hair. Children will have nonred hair when they inherit a dominant nonred hair gene, regardless of whether it is paired with a second nonred hair gene (NN) or a recessive red hair gene (Nr). Red hair can result only from inheriting two recessive genes (rr), which means that both parents must carry the recessive gene for red hair, even if they display nonred hair. Several characteristics are passed through dominant–recessive inheritance (Table 2.1).

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 jointly influence the characteristic (Knopik et al., 2017). For example, consider blood type. Neither the gene for blood type A nor the gene for blood type B dominates. A person with genes blood type A and B will express both and have blood type AB.

Sometimes a gene is stronger than, but does not completely dominate, another gene. In this case some, but not all, characteristics of the recessive gene appear. Individuals who inherit two sickle cell genes develop **sickle cell anemia**, a disorder in which red blood cells become crescent,

FIGURE 2.3 ■ Dominant–Recessive Inheritance

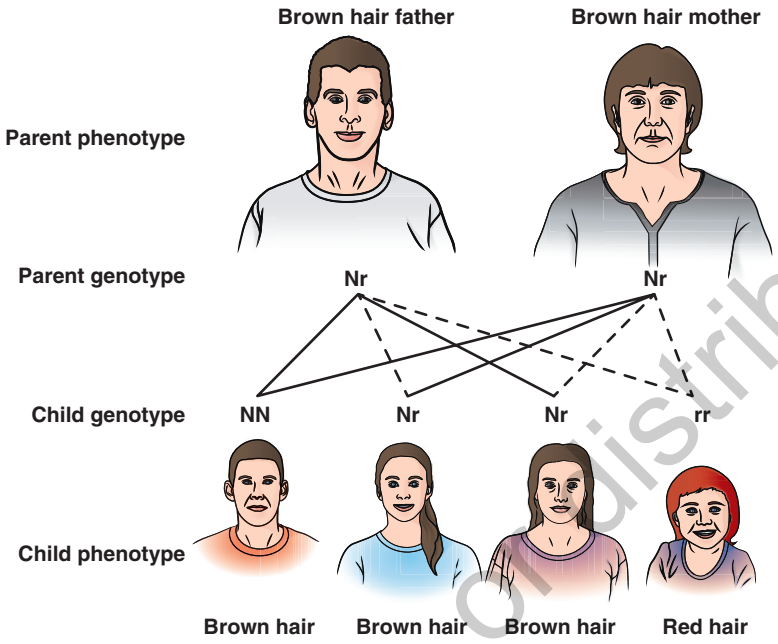


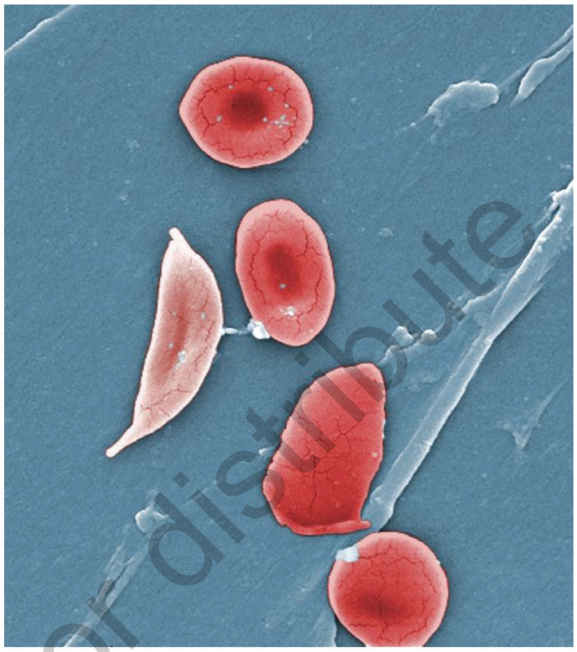
TABLE 2.1 ■ Dominant and Recessive Characteristics

Dominant Trait	Recessive Trait
Dark hair	Blond hair
Curly hair	Straight hair
Hair	Baldness
Nonred 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: Adapted from McKusick-Nathans Institute of Genetic Medicine, 2020.

or sickle, shaped. Cells that are sickle-shaped cannot distribute oxygen effectively throughout the circulatory system and can cause inflammation and damage the blood vessels (Ware et al., 2017). People who carry only one recessive sickle cell gene, do not develop full-blown sickle cell anemia (Chakravorty & Williams, 2015). However, the gene for developing normal blood cells does not completely mask the sickle cell gene. Carriers of the trait for sickle cell anemia

tend to function normally but may show some symptoms such as reduced oxygen distribution throughout the body and exhaustion after exercise (Xu & Thein, 2019). About 5% of African American newborns (and relatively few white or Asian Americans) carry the recessive sickle cell trait (Ojodu et al., 2014).



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.

Genomic Imprinting

The principles of dominant–recessive and incomplete dominance inheritance can account for over 1,000 human traits (Finegold, 2019). But a few traits are determined by **genomic imprinting**, in which the expression of a gene is determined by whether it is inherited from the biological mother or father (Kelly & Spencer, 2017; Thamban et al., 2020). Prader-Willi syndrome and Angelman syndrome are both caused by an abnormality in the 15th chromosome (Kalsner & Chamberlain, 2015). Individuals who acquire the chromosome 15 abnormality from the biological father develop Prader-Willi syndrome (Figure 2.4), a set of specific physical and behavioral characteristics including obesity, insatiable hunger, short stature, motor slowness, and mild to moderate developmental delays (Butler et al., 2016).

When the abnormal chromosome 15 arises from the mother, individuals instead develop Angelman syndrome, characterized by hyperactivity, thin body frame, seizures, disturbances in gait, severe developmental delay or intellectual disability, and speech impairment (Buiting et al., 2016; Dagli & Williams, 2017). Prader-Willi and Angelman syndromes are rare, occurring on average in 1 in 12,000–20,000 persons (Kalsner & Chamberlain, 2015; Spruyt et al., 2018).

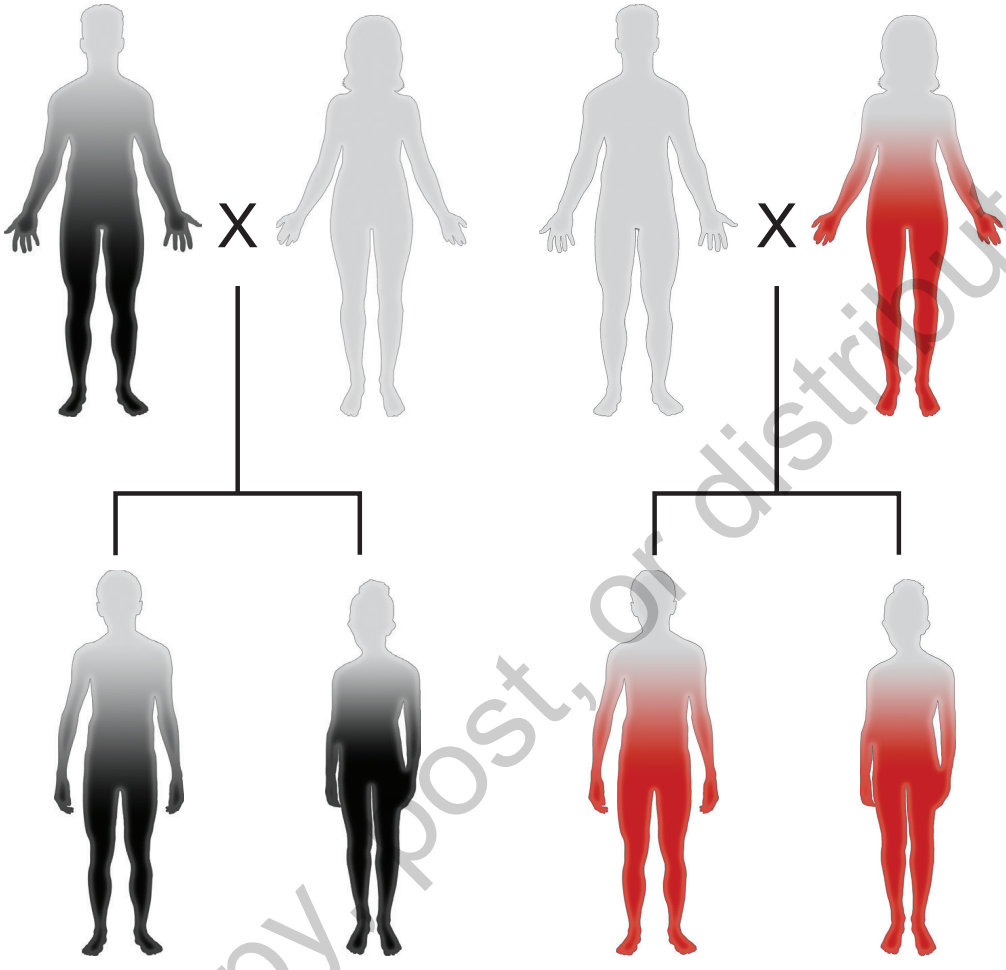
Polygenic Inheritance

Despite our discussion of inheritance thus far, most traits result from the interaction of many genes, known as **polygenic inheritance** (Armstrong-Carter et al., 2021). Hereditary influences act in complex ways, and researchers cannot trace most characteristics to only one or two genes. Examples of polygenic traits include height, intelligence, personality, and susceptibility to certain forms of cancer (Flint et al., 2020). 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. Patterns of genetic inheritance are defined in Table 2.2.

Thinking in Context

1. From an evolutionary developmental perspective (Chapter 1), why might twins occur? 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 traits?
3. Do you think that you might be a carrier of recessive traits? Why or why not?

FIGURE 2.4 ■ Genomic Imprinting



Source: Adapted from C. C. Martin. (1998, July 14). Genomic imprinting. In L. Browder & L. Iten (Eds.), *Dynamic development*.

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. 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.

CHROMOSOMAL AND GENETIC ABNORMALITIES

Many disorders are passed through genetic inheritance, the result of chromosomal abnormalities. Hereditary and chromosomal abnormalities can often be diagnosed prenatally. Others are evident at birth or can be detected soon after an infant begins to develop. Some are discovered only after many years or not at all.

Genetic Disorders

Disorders and abnormalities that are inherited through the parents' genes are passed through the inheritance processes that we have discussed. These include well-known conditions as sickle cell anemia, as well as others that are rare. Some are highly visible and others go unnoticed throughout an individual's life.

Dominant–Recessive Disorders

When genes are passed through dominant–recessive patterns, dominant genes are always expressed and recessive genes are expressed only if paired with another recessive gene. **Phenylketonuria (PKU)** is a common recessive disorder that prevents the body from producing an enzyme that breaks down phenylalanine, an amino acid, from proteins (McKusick-Nathans Institute of Genetic Medicine, 2020). Without treatment the phenylalanine builds up quickly to toxic levels that damage the central nervous system, leading to intellectual disability by 1 year of age. The United States and Canada require all newborns to be screened for PKU (Camp et al., 2014).

PKU illustrates how genes interact with the environment to produce developmental outcomes. Intellectual disability results from the interaction of the genetic predisposition for PKU and exposure to phenylalanine in the diet (Blau, 2016). Infants with PKU are placed on a diet low in phenylalanine, but it is difficult to remove nearly all phenylalanine from the diet. When individuals with PKU maintain a strict diet, they usually attain average levels of intelligence, though they usually score lower than those without PKU (Hofman et al., 2018; Romani et al., 2017). Some cognitive and psychological problems may appear in childhood and persist into adulthood, including poor attention, planning skills, and emotional regulation, as well as depression and anxiety (Christ et al., 2020; Erlich, 2019; Ford et al., 2018; Hawks et al., 2018; Jahja et al., 2017). Common diseases inherited through dominant-recessive inheritance are summarized in Table 2.3.

X-Linked Disorders

A special instance of the dominant–recessive pattern occurs with genes that are located on the X chromosome (Shah et al., 2017). Some recessive genetic disorders, like the gene for red–green color blindness, are carried on the X-chromosome (Table 2.4). Males carry an X and Y chromosome. Because they have only one X chromosome, 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 thereby 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.

Fragile X syndrome is an example of a dominant–recessive disorder carried on the X chromosome (Salcedo-Arellano et al., 2020). Because the gene is dominant, it need appear on only one X chromosome to be displayed by both males and females. Children with fragile X syndrome tend to show moderate to severe intellectual disability and problems with executive function

TABLE 2.3 ■ Diseases Inherited Through Dominant–Recessive Inheritance

Disease	Occurrence	Mode of Inheritance	Description	Treatment
Huntington 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 10,000–15,000	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 3,600–4,000 descendants of Central and Eastern European Jews	Recessive	Degenerative brain disease	None; most die by 4 years of age

Source: Adapted from McKusick-Nathans Institute of Genetic Medicine, 2020.

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.

Chicago Tribune/Contributor/Getty Images

(Raspa et al., 2017; Schmitt et al., 2019). Cardiac abnormalities are common as well as several behavioral mannerisms, including poor eye contact and repetitive behaviors such as hand flapping, hand biting, and mimicking others, behaviors common in individuals with autistic spectrum disorders (Hagerman et al., 2017; Salcedo-Arellano et al., 2020). Fragile X syndrome is often codiagnosed with autism with estimates of about 40% to 60% of boys and 16% to 20% of girls with fragile X syndrome meeting the diagnostic criteria for autism (Bagni & Zukin, 2019; Kaufmann et al., 2017). As carriers, females may show some characteristics of the disorder but tend to display average levels of intelligence.

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 4,000 males and 1 in 8,000 females	Symptoms include cognitive impairment; attention problems; anxiety; unstable mood; long face; large ears; flat feet; and hyper extensible 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: Adapted from McKusick-Nathans Institute of Genetic Medicine, 2020.

Hemophilia, a condition in which the blood does not clot normally, is another recessive disease inherited through genes on the X chromosome (McKusick-Nathans Institute of Genetic Medicine, 2020; Shah et al., 2017). Males with the hemophilia gene display the disorder because the Y chromosome does not have the corresponding genetic information to counter the gene. Females who inherit the gene for hemophilia typically do not show the disorder because the gene on their second X chromosome promotes normal blood clotting and is a dominant gene (d'Oiron, 2019). Females, therefore, can carry the gene for hemophilia without exhibiting the disorder. A female carrier has a 50/50 chance of transmitting the gene to each child.

Chromosomal Abnormalities

Chromosomal abnormalities are the result of errors during cell reproduction, meiosis or mitosis, or damage caused afterward.

Down Syndrome

Occurring in about 1 of about every 1,500 births, trisomy 21, more commonly called **Down syndrome** occurs when a third chromosome appears alongside the 21st pair of chromosomes (de Graaf et al., 2017; McKusick-Nathans Institute of Genetic Medicine, 2020). Down syndrome is associated with marked physical, health, and cognitive attributes, including a short, stocky build and often a round face, almond-shaped eyes, and a flattened nose. Children with Down syndrome tend to show delays in physical and motor development relative to other children, and health problems, such as including congenital heart abnormalities, vision impairments, poor hearing, and immune system deficiencies (Bull, 2020; Diamandopoulos & Green, 2018).

Down syndrome is the most common genetic cause of intellectual disability, but children vary in their abilities (Vissers et al., 2016). Generally, individuals with Down syndrome show greater strengths in nonverbal learning and memory relative to their verbal skills (Grieco et al., 2015).

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.

Halfpoint Images/Getty Images

Expressive language is delayed relative to comprehension. Infants and 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 (Antonarakis et al., 2020; Bull, 2020).

Advances in medicine have addressed many of the physical health problems associated with Down syndrome so that today, the average life expectancy is 60 years of age, as compared with about 25 in the 1980s (Glasson et al., 2014; National Association for Down Syndrome, 2020). Many individuals live into their 70s and 80s. However, Down syndrome is associated with premature aging and an accelerated decline of cognitive functioning (Covelli et al., 2016; Hithersay et al., 2017). Individuals with Down syndrome are at risk to show signs of Alzheimer's disease very early relative to other adults (see Chapter 4) (Antonarakis et al., 2020; Tramutola et al., 2020). 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 (Handen, 2020; N.-C. Lee et al., 2017).

Sex Chromosome Abnormalities

Some chromosomal abnormalities concern the 23rd pair of chromosomes: the sex chromosomes. These abnormalities result from either an additional or missing sex chromosome. Given their different genetic makeup, sex chromosome abnormalities yield different effects in males and females (Table 2.5)

One of the most common sex chromosome abnormalities is **Klinefelter syndrome**, in which males are born with an extra X chromosome (XXY) (McKusick-Nathans Institute of Genetic Medicine, 2020; Wistuba et al., 2017). Symptoms range in severity, but most men are unaware of the disorder until they are tested for infertility (Bird & Hurren, 2016; Gravholt et al., 2018). Severe symptoms include 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 (Bonomi et al., 2017). As adults, men with Klinefelter syndrome are at risk for a variety of disorders that are more common in women, such as osteoporosis (Juul et al., 2011).

The sex chromosome abnormality known as **Turner syndrome** occurs when a female is born with only one X chromosome (McKusick-Nathans Institute of Genetic Medicine, 2020). Girls with Turner syndrome show abnormal growth patterns. Their ovaries do not develop normally, they do not ovulate, and they are infertile (Culen et al., 2017; Davis et al., 2020). As adults, they are short in stature and have webbed necks (extra folds of skin) (Gravholt et al., 2019). Malformations of the heart, diabetes, autoimmune disorders, and early osteoporosis are common (Gravholt et al., 2019). Children with Turner syndrome may show difficulty with

TABLE 2.5 ■ Sex Chromosome Abnormalities

Female Genotype	Syndrome	Description	Prevalence
XO	Turner	Abnormal growth patterns, delayed puberty, lack prominent female secondary sex characteristics, and infertility. Short adult stature, webbing around their neck.	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. Many cases go unnoticed.
Male Genotype	**Syndrome**	**Description**	**Prevalence**
XXY	Klinefelter	High-pitched voice, short stature, feminine body shape, and infertility. Increased risk for osteoporosis and other disorders that are more common in women.	1 in 1,000 males
XYY	Jacob's	Accompanied by high levels of testosterone.	Unknown. Many cases go unnoticed.

Source: Adapted from McKusick-Nathans Institute of Genetic Medicine, 2020.

visual-spatial reasoning and memory, attention, executive functioning, motor skills, and math skills (Hutaff-Lee et al., 2019). They are also prone to social difficulties, anxiety, and depression (Christopoulos et al., 2008; Powell & Schulte, 2011). If diagnosed early, regular injections of human growth hormones can increase stature and can result in breast growth and menstruation (Culen et al., 2017; Klein et al., 2020).

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. A mutation may involve only one gene or many genes. It is estimated that as many as one half of all conceptions include mutated chromosomes (Taneri et al., 2020). Most mutations are fatal—the developing organism often dies very soon after conception, often before the woman knows she is pregnant (Sadler, 2018).

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. The sickle cell gene (discussed earlier in this chapter) is a mutation that originated in areas where malaria is widespread, such as Africa (Ware et al., 2017) and serves a protective role against malaria (Uyoga et al., 2019).

Children who inherited a single sickle cell gene were more resistant to malarial infection and more likely to survive and pass it along to their offspring (Croke et al., 2017; Gong et al., 2013). The sickle cell gene is not helpful in places of the world where malaria is not a risk. It is becoming less frequent in places in the world where malaria is uncommon. For example, only 5% to

8% of African Americans are carriers, compared with as much as 30% of Black Africans in some African countries (Maakaron et al., 2012; Ojodu et al., 2014) Therefore, the developmental implications of genotypes—and mutations—are context-specific, posing benefits in some contexts and risks in others.

Assisted Reproductive Technology

Nearly 2% of infants in the United States are conceived through **assisted reproductive technology (ART)**, alternative methods of conception that rely on medical technology (Centers for Disease Control and Prevention, 2020a). Candidates for ART include individuals and couples at risk for bearing children with genetic or chromosomal abnormalities, couples who experience infertility, and single adults and gay and lesbian couples who wish to conceive.

There are racial, ethnic, and socioeconomic disparities in the use of ART. White, Asian American, college-educated, and high socioeconomic status (SES) women are more likely to give birth with ART than are Black and Hispanic women (Janitz et al., 2016; Tierney & Cai, 2019). Race and ethnicity are often linked with SES and disparities in health care in the United States. Socioeconomic factors play a large role in access to infertility treatment and reproductive technology (Dieke et al., 2017).

Artificial Insemination

The simplest, least invasive type of alternative conception is **artificial insemination**, the injection of sperm into a woman. Sperm is obtained from a partner or donor. Artificial insemination is the least expensive alternative method of conception, but the success rate is low, usually requiring multiple cycles. The injection costs range from about $300 to $1,000 per cycle (Harris, 2020). Women and couples who seek donor sperm may also expect to pay about $700 to $1,000 per vial.

In vitro Fertilization

In contrast with artificial insemination, where conception occurs inside of the women's body, **in vitro fertilization** initiates conception outside of the woman's body. A woman is prescribed hormones that stimulate the maturation of several ova, which are surgically removed. The ova are placed in a dish and sperm are added. One or more ova are fertilized, and the resulting cell begins to divide. After several cell divisions, the cluster of cells are placed in the woman's uterus. If they implant into the uterus and begin to divide, a pregnancy has occurred.

The success rate of in vitro fertilization is about 50% and varies with the mother's age (Centers for Disease Control and & Prevention, 2019b). For instance, the percentage of embryo transfers resulting in live births is 53% for 35- to 37-year-old women, 36% in 38- to 40-year-old women, 25% in 41- and 42-year-old women, and only 11% in women over age 43. In vitro fertilization is expensive, costing an average of over $12,400 per trial, not including medication, and often requires multiple cycles, posing a financial burden too great for low SES women and couples (Asch & Marmor, 2020; Teoh & Maheshwari, 2014).

Infants conceived by in vitro fertilization show no differences in growth, health, development, and cognitive function relative to infants conceived naturally (Farhi et al., 2019; Fauser et al., 2014). Because in vitro fertilization permits cells to be screened for genetic problems prior to implantation, in vitro infants are not at higher risk of congenital (birth) abnormalities (Fauser et al., 2014). Yet over one third of births from artificial insemination include more than one infant (Sunderam et al., 2019). Multiple gestations increase risk for low birthweight, prematurity, and other poor outcomes (Sullivan-Pyke et al., 2017).

Surrogacy

Surrogacy is an alternative form of reproduction known in which a woman (the surrogate) is impregnated and carries a fetus to term and agrees to turn the baby over to a woman, man, or couple who will raise it. Single parents, same-sex couples, and couples in which one or both members are infertile choose surrogacy. Sometimes the surrogate carries a zygote composed of one or both of the couple's gametes. Other times, the ova, sperm, or zygote are donated. Despite several highly publicized cases of surrogate mothers deciding not to relinquish the infant, most surrogacies are successful.

Roughly 3,000 babies are born through surrogacy in the United States each year (Beitsch, 2017). Longitudinal research suggests no psychological differences through age 14 between children born through surrogacy compared with other methods, including children born to gay father and lesbian mother families (Carone et al., 2018, 2020; Golombok, 2013; Golombok et al., 2017). In addition, women who raise children conceived through surrogacy are similar to those who raise birthed children, with no negative effects (Jadva et al., 2015; Söderström-Anttila et al., 2015).

We have seen that reproductive technology is expensive. Surrogacy is often prohibitively expensive for most prospective parents, limiting its access to parents with high SES. Prospective parents pay for the surrogate's medical care, attorney, travel expenses, health care, and more, which can amount to $100,000 or more (Caron, 2020). Finally, surrogacy may pose ethical issues. Carrying a fetus to term poses physical and mental health risks to the surrogate. Relinquishing a newborn is difficult, even with fore planning, posing emotional risks to the surrogate. The financial incentives to surrogate a fetus are substantial. Although paying a surrogate is illegal, women are often compensated for the physical and emotional burden of surrogating a fetus. A surrogate tends to receive at least $30,000 to $55,000 to surrogate a fetus, sums that may be difficult for low SES women to resist (Beitsch, 2017; Harrison, 2017)

Thinking in Context

1. Recall from Chapter 1 that most developmental scientists agree that nature (genetics) and nurture (environment) interact to influence development. Choose a genetic or chromosomal disorder discussed in this section and explain how it illustrates the interaction of genes and context.
2. Assisted reproductive technology is not easily available to all individuals and couples. What are some of the barriers to obtaining assisted reproductive technology? Which are most important? Should everyone have equal access to these technologies? What are challenges to improving access?
3. Provide advice to Eduardo and Natia, a couple in their mid-30s who are seeking reproductive assistance. What are their options, and what are the advantages and disadvantages of each option?

HEREDITY AND ENVIRONMENT

We have seen that our genotype, inherited from our biological parents, influences all of our observable traits, from hair and eye color to personality, health, and behavior. However, genotypes alone do not determine our **phenotype**, the traits and characteristics we display. Phenotypes result from the interaction of genotypes and our experiences.

Methods of Behavior Genetics

Behavior genetics is the study of how genes and experience combine to influence the diversity of human traits, abilities, and behaviors (Knopik et al., 2017; Plomin, 2019). Even traits with a strong genetic component, such as height, are modified by environmental influences (Jelenkovic et al., 2016). Most human traits, such as intelligence, are influenced by multiple genes, which have multiple variants that can each interact with the environment in different ways (Armstrong-Carter et al., 2021; Briley et al., 2019; Plomin et al., 2016).

Behavior geneticists seek to estimate 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 in phenotypes is a result of the environment and experiences (Fowler-Finn & Boutwell, 2019; Nivard et al., 2017).

Behavior geneticists conduct twin studies and adoption studies to compare people who live together and share varying degrees of relatedness (York, 2020). *Twin studies* compare identical and fraternal twins to estimate how much of a trait or behavior is attributable to genes. Recall that identical (monozygotic) twins share 100% of their genes because they originated from the same zygote. Like all nontwin siblings, fraternal (dizygotic) twins share 50% of their genes as they resulted from two different fertilized ova and two genetically different zygotes. If genes affect a given attribute, identical twins should be more similar than fraternal twins because identical twins share 100% of their genes, whereas fraternal twins share about half. 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.

Adoption studies 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 an environment but not genes (York, 2020). 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. The similarities are influenced by the environment if the children are more similar to their adoptive parents (and adoptive siblings).

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 intelligence scores than do fraternal twins (Plomin, 2019). A classic 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 2.6 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, temperament, emotionality, and susceptibility to various illnesses such as obesity, heart disease and cancer, anxiety, poor mental health, and a propensity to be physically aggressive (Bralten et al., 2019; Goodarzi, 2018; Morneau-Vaillancourt et al., 2019; Purves et al., 2019; Trucco et al., 2018). Yet even traits that are thought to be heavily influenced by genetics can be modified by physical and social interventions. 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; Jelenkovic et al., 2016). Even

TABLE 2.6 ■ 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, T. J., & McGue, M. (1981). Familial studies of intelligence: A review. *Science, 212*(4498), 1055–1059.

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

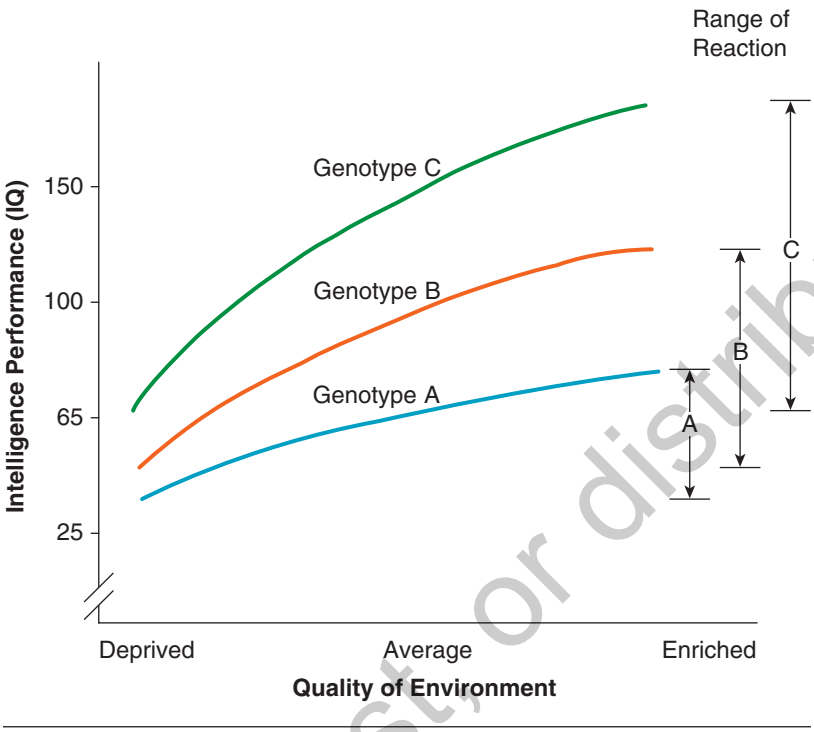
Genes and the environment work together in complex ways to determine our characteristics, behavior, development, and health (Morgan et al., 2020; Ritz et al., 2017). **Gene–environment interactions** refer to the dynamic interplay between our genes and our environment. Several principles illustrate these interactions.

Range of Reaction

The effects of the environment varies with the genetic makeup of the individual (Briley et al., 2019). 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** (Figure 2.5), a wide range of potential expressions of a genetic trait, depending on environmental opportunities and constraints (Gottlieb, 2007).

Consider height. Height is largely a function of genetics, yet an individual may show a range of sizes depending on environment and behavior (Jelenkovic et al., 2016). Children born to two very tall parents may have the genes to be tall. But unless they have adequate nutrition, they will not fulfil their 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 fulfil 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; Morgan et al., 2020). In this way, genetics sets the range of development outcomes and the environment influences where, within the range, that person will fall. Gene–environment interactions are complex and often difficult to predict, partly because individuals vary in their sensitivity to environmental stimuli (Belsky & Hartman, 2014). Some children may be more affected by environmental stimuli due to their genetic makeup (Briley et al., 2019).

FIGURE 2.5 ■ Range of Reaction



Source: Adapted from Gottesman, I. I. (1963). Heritability of personality. *Psychological Monographs: General and Applied, 77*, 1–21.

Canalization

Some traits have a narrow reaction range. These traits illustrate **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; Posadas & Carthew, 2014; Takahashi, 2019). 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 (Adolph & Franchak, 2017). For example, children reared in impoverished international orphanages and exposed to extreme environmental deprivation demonstrated delayed motor development, with infants walking 5 months to 1 year later than expected (Chaibal et al., 2016; L. Miller et al., 2008).

Motor development is not entirely canalized, however, because some minor changes in the environment can subtly alter its pace and timing. 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 (Adolph & Hoch, 2019). 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 Correlation

Heredity and environment are each a powerful influence on development. Not only do they interact, but environmental factors often support hereditary traits (Briley et al., 2019; Saltz,

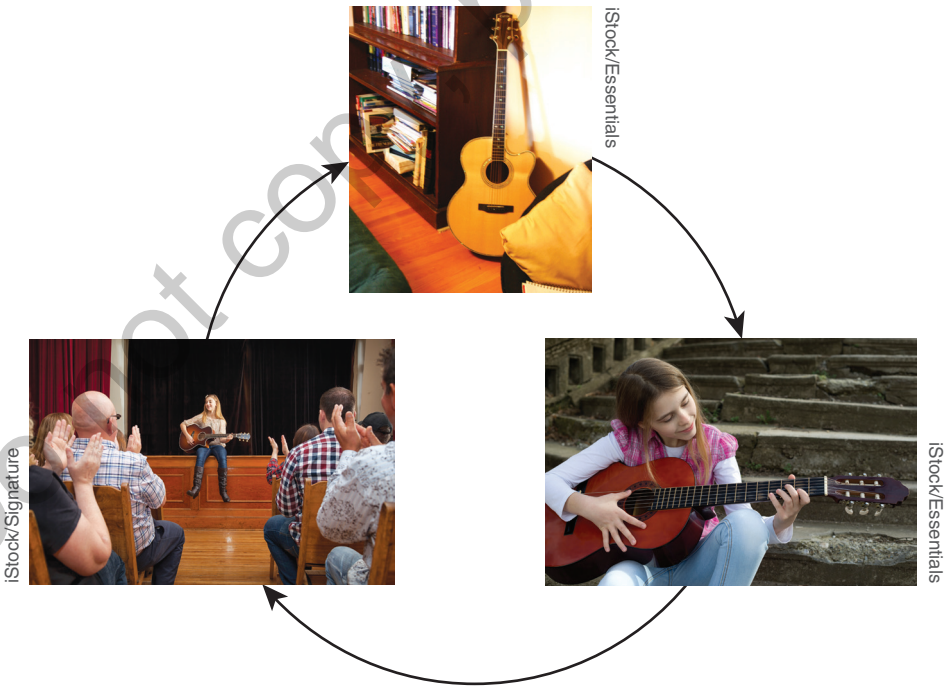
2019; Scarr & McCartney, 1983). **Gene–environment correlation** refers to the finding that many genetically influenced traits tend to be associated with environmental factors that promote their development (K. Lynch, 2016). That is, genetic traits influence children's behavior, which is often supported or encouraged by the environment (Knafo & Jaffee, 2013). There are three types of gene–environment correlations: passive, evocative, and active.

Passive gene–environment correlation. Parents create homes that reflect their own genotypes. Because parents are genetically similar to their children, the homes that parents create support their own preferences but they also correspond to their child's genotype—an example of a *passive gene–environment correlation* (Wilkinson et al., 2013). It is a passive gene–environment correlation because it occurs regardless of the child's behavior. For example, parents might provide genes that predispose a child to develop music ability and create a home environment that supports the development of music ability, such as by playing music in the home and owning musical instruments (Corrigall & Schellenberg, 2015)(Figure 2.6). This type of gene–environment correlation tends to occur early in life because parents create rearing environments for their infants and young children.

Evocative gene–environment correlation. 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 environments,

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).



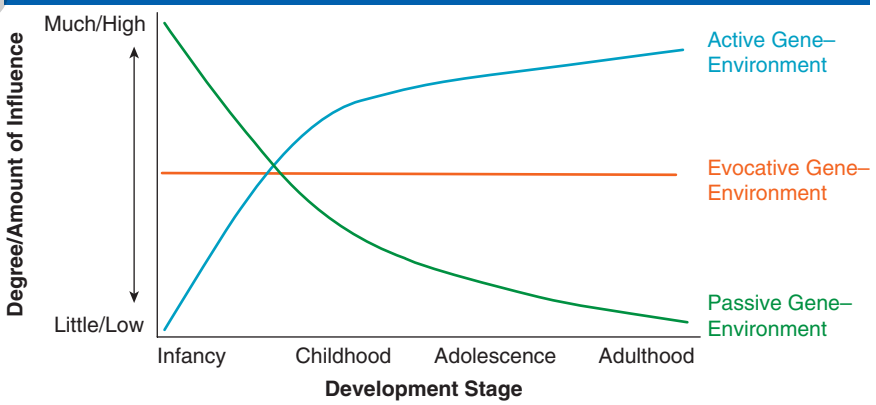
Source: Images from iStock/Signature; iStock/Essentials.

which shape development in ways that support the genetic trait (Pieters et al., 2015; Saltz, 2019). 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 and affection from others, including their parents, which in turn supports the tendency to be cheerful (Klahr et al., 2013). In this way, genotypes influence the physical and social environment to respond in ways that support the genotype. To return to the music example, a child with a genetic trait for music talent will evoke pleasurable responses (e.g., parental approval) when the child plays music; this environmental support, in turn, encourages further development of the child's musical trait.

Active gene–environment correlation. Children also take a hands-on role in shaping their development. Recall from Chapter 1 that individuals are active in their development. 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 or her genetic predisposition. 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 (Corrigall & Schellenberg, 2015). This tendency to actively seek out experiences and environments compatible and supportive of our genetic tendencies is called **niche picking** (Saltz, 2019; Scarr & McCartney, 1983).

The strength of passive, evocative, and active gene–environment correlations changes with development, as shown in Figure 2.7 (K. Lynch, 2016; 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. 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

FIGURE 2.7 ■ Gene–Environment Correlations Over the Lifespan



fit their genetic propensities. As they age, identical twins—even those reared apart—become alike in attitudes, personality, cognitive ability, strength, mental health, and preferences; they also tend to select similar spouses and best friends (McGue & Christensen, 2013; Plomin & Von Stumm, 2018; York, 2020).

Gene–Environment (G x E) Interactions

We have seen that behavior is influenced by gene–environment interactions. Genes may provide a reaction range through which environmental factors act. Some genes severely limit developmental outcomes (canalization). Just as some genes might increase our susceptibility to environmental risks, others might increase our sensitivity to, and the effectiveness of, environmental interventions (Bakermans-Kranenburg & van IJzendoorn, 2015; Chhangur et al., 2017). The effects of genes vary with environmental influences and not all genotypes respond to environmental influences in the same way (Fowler-Finn & Boutwell, 2019). Although behavior geneticists have learned a great deal about genetic influences on behavior, effects are often unpredictable (Flint et al., 2020).

Consider the following classic study. Caspi and colleagues (2002) followed a sample of boys from birth until adulthood. Although children who experience child maltreatment, or abuse, tend to show developmental and behavioral problems, the effects of maltreatment varied among the boys. Upon further study the researchers were surprised to find that the link between maltreatment and violence varied with the gene that controls monoamine oxidase A (MAOA), an enzyme that regulates specific chemicals in the brain. Only boys who carried the gene for low levels of MAOA were at risk for becoming violent after experiencing maltreatment.

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 nonmaltreated 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 experienced abuse early in life. These findings have been replicated in another 30-year longitudinal study of boys (Fergusson et al., 2011) as well as a meta-analysis of 27 studies (Byrd & Manuck, 2014).

Similar findings of a MAOA gene × 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 et al., 2007; Manuck & McCaffery, 2014; Nikulina et al., 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 × environment interaction on emotional reactivity and aggression but to a much lesser extent than boys, whereas other studies suggest no relationship (Byrd et al., 2018; Byrd & Manuck, 2014).

The MAOA gene influences adaptation to adversity, such as the trauma of child maltreatment.

iStock/Juanmonino

Behavioral genetics research has yielded fascinating results. However, the conclusions of these studies pertain to populations, groups of people, not individuals. Findings from behavior genetic research cannot predict individual behavior (Turkheimer, 2019). A final important criticism of behavior genetic research is that, like many other areas of research, its samples are not diverse. Ethnically diverse samples and those of low SES are underrepresented, limiting conclusions (Sirugo et al., 2019).

Epigenetic Framework

Development is the product of a dynamic interaction of biological and contextual forces. Genes provide a blueprint for development, but phenotypic outcomes (individuals' characteristics) are not predetermined; rather, they vary with environmental factors. Recently scientists have determined that environmental factors do not simply interact with genes to determine people's traits, but they can determine *how* genes are expressed through a process known as **epigenetics** (Carlberg & Molnar, 2019; D. S. Moore, 2017).

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, whether they are turned on or off. The epigenome carries the instructions that determine what each cell in your body will become, whether heart cell, muscle cell, or brain cell. Those instructions are carried out by turning genes on and off.

At birth, each cell in our body turns on only a fraction of its genes. The epigenome instructs genes to be turned on and off over the course of development and also in response to the environment (Meaney, 2017). Epigenetic mechanisms determine how genetic instructions are carried out to determine the phenotype (Lester et al., 2016; Pinel et al., 2018). Environmental factors such as toxins, injuries, crowding, diet, and responsive parenting can influence the expression of genetic traits by determining what genes are turned on and off (O'Donnell & Meaney, 2020). In this way, even traits that are highly canalized can be influenced by the environment.

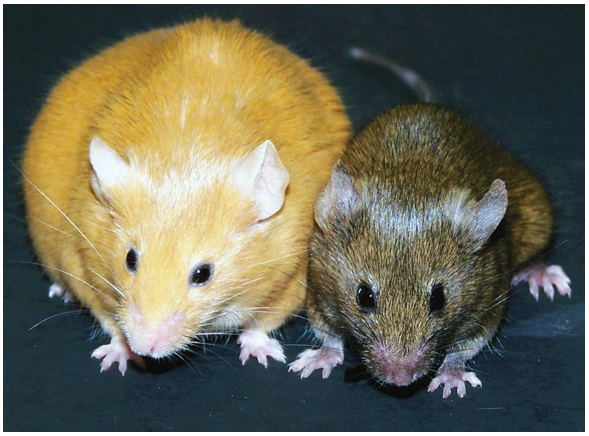
Epigenetic Processes in Animals

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, are shaped much like a pincushion, and are 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). Both of the mice in Figure 2.8 carry the agouti gene, yet they look very different. The brown mouse is slender, is lean, and has a low risk of developing diabetes and cancer, living well into old age. Why are these mice so different? Epigenetics. 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 Jertle discovered that the pregnant 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.

FIGURE 2.8 ■ Agouti Gene

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 and in the brown mouse it is turned off.



Source: Wikipedia/Randy Jirtle and Dana Dolinoy/Creative Commons 3.0

Epigenetic Processes in People

Epigenetic processes also influence human development. Consider brain development (O'Donnell & Meaney, 2020). Providing infants with a healthy diet and opportunities to explore the world will support the development of brain cells, governed by epigenetic mechanisms that switch genes on and off. Conversely, epigenetic changes that accompany exposure to toxins or extreme trauma might suppress the activity of some genes, potentially negatively influencing brain development. In this way, individuals' neurological capacities are the result of epigenetic interactions among genes and contextual factors (Lerner & Overton, 2017) (Figure 2.9). 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 (Lickliter & Witherington, 2017).

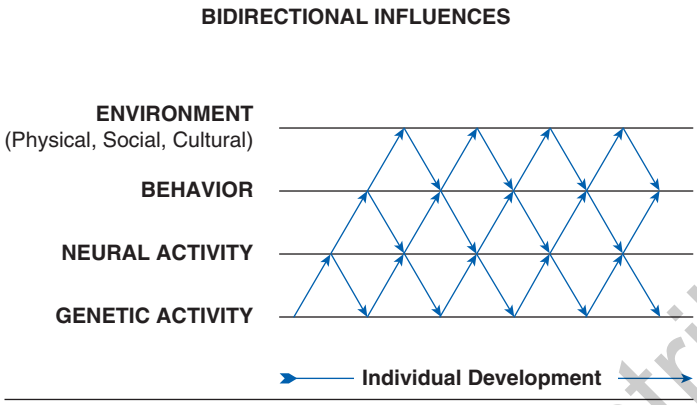
The epigenome can be influenced by the environment before birth, after birth, and can even be passed by males and females from one generation to the next (Legoff et al., 2019; Szyf, 2015). This means that what you eat and do today could affect the epigenome—the development, characteristics, and health—of your children, grandchildren, and great-grandchildren (Bošković & Rando, 2018; Grover & Jenkins, 2020; Vanhees et al., 2014).

Thinking in Context

Describe a skill or ability in which you excel. How might your ability be influenced by your genes and your context?

1. Identify a passive gene–environment correlation that may contribute to your ability. How has your environment influenced your ability?
2. Provide an example of an evocative gene–environment correlation. How have you evoked responses from your context that influenced your ability?
3. Explain how your ability might reflect an active gene–environment correlation.
4. What of these types of gene–environment correlations do you think best accounts for your ability? Why?

FIGURE 2.9 ■ Epigenetic Framework



Source: Adapted from Gottlieb, G. (2007). Probabilistic epigenesis. *Developmental Science, 10*(1), 1–11

PRENATAL DEVELOPMENT

Conception, the union of ovum and sperm, marks the beginning of prenatal development. Over the next 38 weeks, the human progresses from fertilization to birth. During this transformation the zygote undergoes several periods of development, finally emerging from the womb as a neonate.

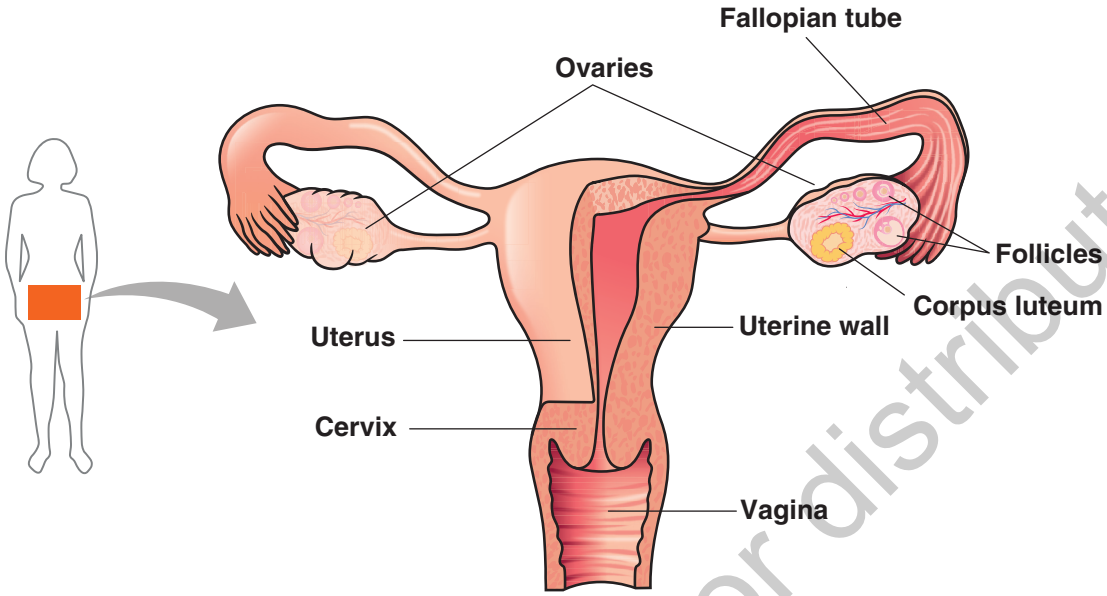
Conception

A woman can conceive only during a short window of time each month. About every 28 days, an ovum bursts from one of the ovaries into the long, thin fallopian tube that leads to the uterus; this event is known as ovulation (Figure 2.10). The ovum is the largest cell in the human body, yet it is only 1/175th of an inch in diameter (about the size of the period at the end of this sentence). Over several days, the ovum travels down the fallopian tube, which connects the ovaries to the uterus, while the corpus luteum, the spot on the ovary from which the ovum was released, secretes hormones that cause the lining of the uterus to thicken in preparation for the fertilized ovum (Sadler, 2018). If fertilization does not occur, the uterus lining is shed through menstruation about 2 weeks after ovulation.

Each day, a man's testes produce millions of sperm composed of a pointed head packed with 23 chromosomes' worth of genetic material and a long tail. During ejaculation, about 360 million, and as many as 500 million, sperm are released, bathed in a protective fluid called semen (K. L. Moore et al., 2019). After entering the female's vagina, sperm travel through the cervix into the uterus and onward to the fallopian tube, where an ovum may be present. The journey is difficult: Some sperm get tangled up with other sperm, some travel up the wrong fallopian tube, and others do not swim vigorously enough to reach the ovum. On average, about 300 sperm reach the ovum, if one is present (Webster et al., 2018). Those that travel up the fallopian tube can live up to 6 days, able to fertilize a yet unreleased ovum. The ovum remains viable for about only a day after being released into the fallopian tube.

When a sperm penetrates the ovum, its tail falls off, and the sperm's genetic contents merge with that of the ovum. At the moment of conception, the zygote contains 46 chromosomes, half from the ovum and half from the sperm. After fertilization, the zygote rapidly transforms into a multicelled organism. Prenatal development takes place over three developmental periods: (1) the germinal period, (2) the embryonic period, and (3) the fetal period.

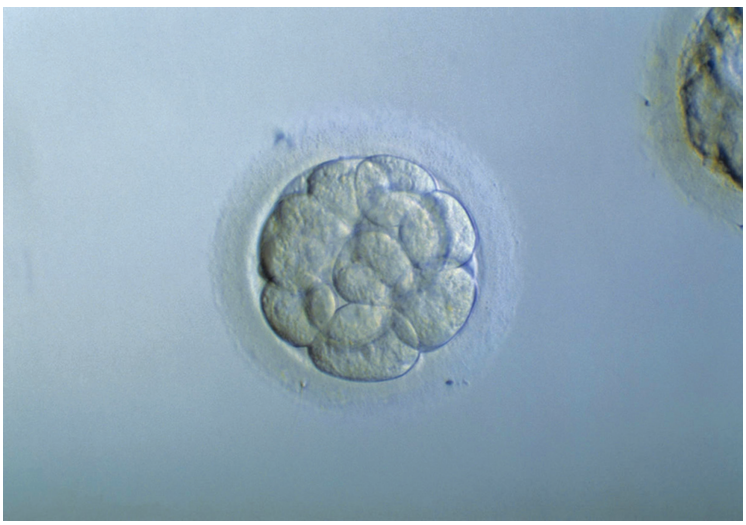
FIGURE 2.10 ■ Female Reproductive System



Germinal Period (0 to 2 Weeks)

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 the middle, forming two identical cells (Webster et al., 2018). This process is called cleavage, and it continues at a rapid pace. Each of the two cells splits to form four cells, then eight, and so on (Figure 2.11). Each of the resulting cells is identical until about the third set of cell divisions. This process of cell division continues rapidly. Any of these cells may become a person (and sometimes do, in the case of monozygotic or identical twins).

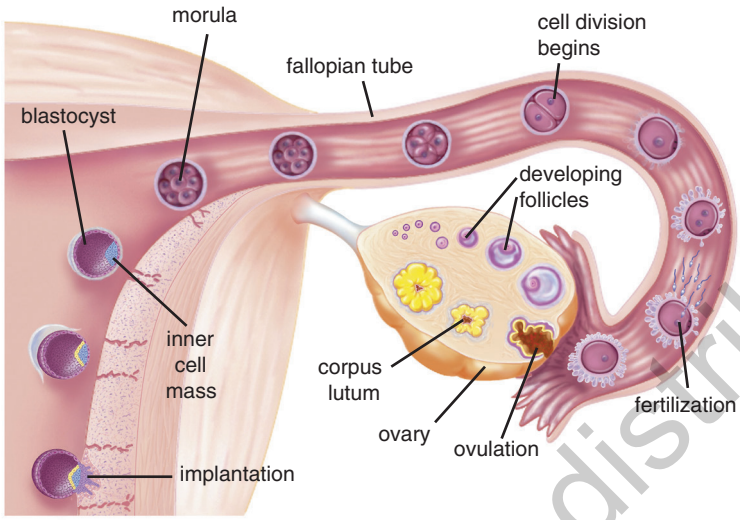
Cell differentiation begins roughly 72 hours after fertilization when the organism consists of about 16 to 32 cells. Differentiation means that the cells begin to specialize and are no longer identical. By 4 days, the organism consists of about 60 to 70 cells formed into a hollow ball called a **blastocyst**, 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 (K. L. Moore et al., 2019).



This ball of cells, known as a morula, is formed at about 3 days after conception. Each of these cells is identical. Differentiation has not yet begun.

Pascal Goetgheluck/Science Source

FIGURE 2.11 ■ The Germinal Period

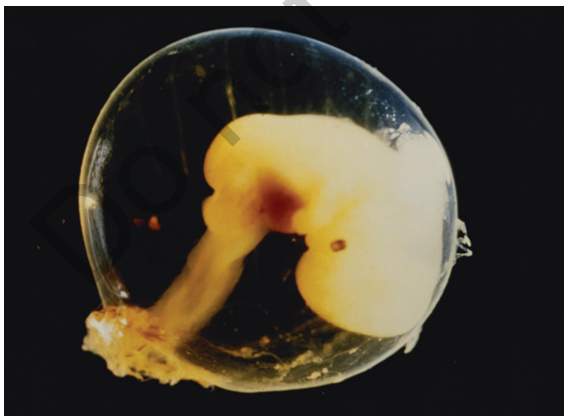


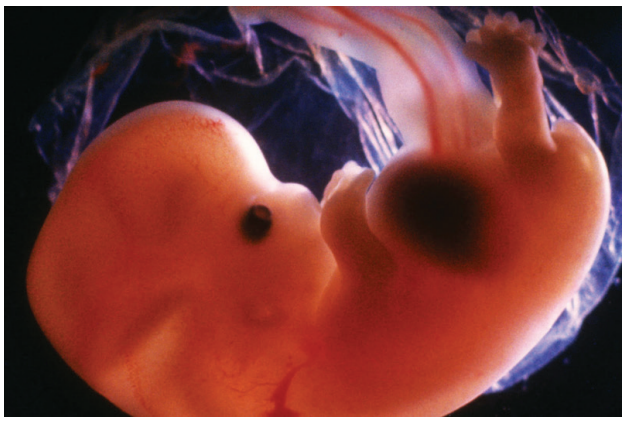
Embryonic Period (3 to 8 Weeks)

After implantation, during the third week after conception, the developing organism, now called an **embryo**, begins the most rapid period of structural development in the lifespan. All of the organs and major body systems form during the embryonic period. The mass of cells composing the *embryonic disk* forms layers, which will develop into all of the body's major organs. The *ectoderm*, the upper layer, will become skin, nails, hair, teeth, sensory organs, and the nervous system. 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.

FIGURE 2.12 ■ The Embryonic Period

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





Source: Petit Format/Science Source

As the embryo develops, support structures form to protect it, provide nourishment, and remove wastes. The **amnion**, a membrane that holds amniotic fluid, surrounds the embryo providing temperature regulation, cushioning, and protection from shocks. The **placenta**, a principal organ of exchange between the mother and developing organism begins to form. It contains tissue from both the mother and embryo and, once formed, it will act as a filter, enabling the exchange of nutrients, oxygen, and wastes through the umbilical cord. The placenta is also a protective barrier, preventing some toxins from entering the embryo's bloodstream as well as keeping the mother and embryo's bloodstreams separate. Still, many toxins can pass through the placenta, including drugs and chemicals, such as alcohol, cannabis, and opioids, as we will discuss later.

About 22 days after conception marks a particularly important change. The ectoderm folds to form the **neural tube**, which will develop into the central nervous system (brain and spinal cord) (Webster et al., 2018). 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. Arm buds appear during days 26 and 27, followed by leg buds on days 28 through 30 (Sadler, 2018). At about this time, a tail-like appendage extends from the spine, disappearing at about 55 days after conception. 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 (K. L. Moore et al., 2019). 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 1 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 problems during the embryonic period often cause a miscarriage, or spontaneous abortion (loss of the embryo); indeed, most miscarriages are the result of chromosomal abnormalities. Organisms with the most severe abnormalities 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 (Chou et al., 2020).

Fetal Period (9 Weeks to Birth)

During the **fetal period**, from the ninth week to birth, the organism, called a **fetus**, grows rapidly, and its organs become more complex and begin to function. Now 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, 2018).

By the 14th week, limb movements are coordinated but are 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. 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 (K. L. Moore et al., 2019). 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. A startle response has been reported at 22 to 23 weeks in response to sudden vibrations and noises (Hepper, 2015). The startle response is a basic reflex controlled by the developing central nervous system and is not a voluntary movement. 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.

During the last 3 months of pregnancy, the fetal body grows substantially in weight and length; 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 (Andescavage et al., 2016). 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.

The expected date of delivery is about 266 days or 38 weeks, from conception (40 weeks from the mother's last menstrual period), but about 1 in every 10 American births is premature (Centers for Disease Control and Prevention, 2019a). 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 (K. L. Moore et al., 2019). Although a 23-week fetus born prematurely may survive in intensive care, its immature respiratory system places it at risk; only about one third of infants born at 23 weeks' gestation survive (Esteves et al., 2016; Stoll et al., 2015). By 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 25 weeks survive, and 94% of those born at 27 weeks also survive (Myrhaug et al., 2019). As we will discuss later in this chapter, premature infants experience heightened risk for short- and long-term impairments and disabilities.

Prenatal Diagnosis

Expectant parents view ultrasound images of the developing fetus.

Jose Luis Pelaez Inc/Getty Images

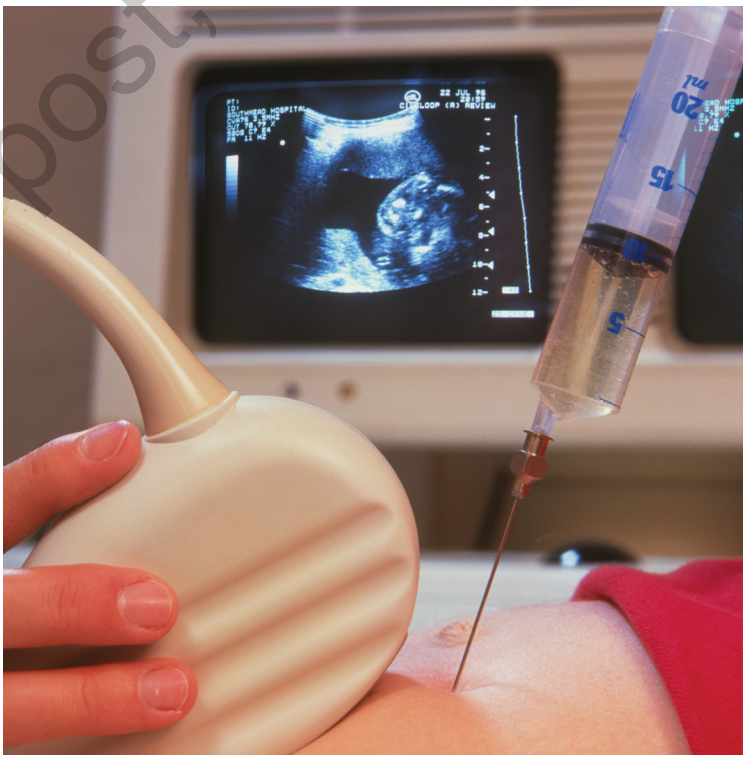
Virtually all pregnant women undergo examinations to determine the health of the fetus. The most widespread and routine diagnostic procedure is **ultrasound**, in which high-frequency sound waves directed at the mother's abdomen provide clear images of the womb represented on a video monitor. Ultrasound enables physicians to observe the fetus, measure fetal growth, judge gestational age, reveal the sex of the fetus, detect multiple pregnancies (twins, triplets, etc.), and determine physical abnormalities in the fetus. At least 80% of women in the United States receive at least one prenatal ultrasound scan (Sadler, 2018). Three to four screenings over the duration of pregnancy are common in order to evaluate fetal development.

Fetal MRI applies MRI technology to image the fetus's body and diagnose malformations (Aertsen et al., 2020). Fetal MRI can detect abnormalities throughout the body, including the central nervous system (Griffiths et al., 2017; Masselli et al., 2020). MRI is safe for mother and fetus in the second and third trimesters but is expensive and has limited availability in some communities (Patenaude et al., 2014)

Amniocentesis is a prenatal diagnostic procedure in which a small sample of the amniotic fluid surrounding the fetus is extracted from the mother's uterus through a long, hollow needle guided by ultrasound as it is inserted into the mother's abdomen (Odibo, 2015). The amniotic fluid contains fetal cells, which are grown in a laboratory dish to create enough cells for genetic analysis. Genetic analysis is then performed to detect genetic and chromosomal anomalies. Amniocentesis is less common than ultrasound, because it poses greater risk to the fetus, but it is safe (Homola & Zimmer, 2019). It is recommended for women aged 35 and over, especially if the woman and partner are both known carriers of genetic diseases (Vink & Quinn, 2018a). Usually amniocentesis is conducted between the 15th and 18th week of pregnancy. Conducted any earlier, an amniocentesis may increase the risk of miscarriage (Akolekar et al., 2015). Test results generally are available about 2 weeks after the procedure because it takes that long for the genetic material to grow and reproduce to the point where it can be analyzed.

Chorionic villus sampling (CVS) also samples genetic material and can be conducted earlier than amniocentesis, usually between 10 and 14 weeks of gestation (Vink & Quinn, 2018b). CVS requires studying a small amount of tissue from the chorion, part of the membrane surrounding the fetus. The tissue sample is obtained through a long needle inserted either abdominally or vaginally, depending on the location of the fetus. Results are available about 1 week following the procedure. CVS is relatively painless, poses few risks to the fetus, and, like amniocentesis, has a 100% diagnostic success rate (Salomon et al., 2019; Shim et al., 2014). CVS should not be conducted prior to 10 weeks gestation because some studies suggest an increased risk of limb abnormalities and miscarriages (Shahbazian et al., 2012).

Noninvasive prenatal testing (NIPT) screens the mother's blood to detect chromosomal abnormalities. Cell-free fetal DNA (chromosome fragments that result from the breakdown of fetal cells) circulates in maternal blood in small concentrations that can be detected and studied by sampling the mother's blood (Hartwig et al., 2017; Warsof et al., 2015). Testing can be done after 10 weeks, typically between 10 and 22 weeks. Given that the test involves drawing blood from the mother, there is no risk to the fetus. The use of NIPT has increased dramatically in the United States and other countries (Hui et al., 2017). NIPT can provide accurate sex determination, but



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.

Saturn Stills/Science Source

NIPT cannot detect as many chromosomal abnormalities as amniocentesis or CVS and is less accurate (Hartwig et al., 2017; Villela et al., 2019). Researchers have identified the entire genome sequence using NIPT, suggesting that someday, NIPT may be as effective as other, more invasive techniques (Tabor et al., 2012). In consultation with their obstetrician, pregnant women and their partners should carefully weigh the risks and benefits of any procedure designed to monitor prenatal development. Table 2.7 summarizes methods of prenatal diagnosis.

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. Suppose that you are a health care provider tasked with explaining prenatal diagnostic choices to a 38-year-old woman pregnant with her first child. How would you explain the various choices? What information would you provide about their purpose and the advantages and disadvantages of each? Which would you advise? Why?

TABLE 2.7 ■ Methods of Prenatal Diagnosis

	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 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 fetal 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)	CVS 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 fetal 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 abnormalities when conducted prior to 10 weeks' gestation.

	Explanation	Advantages	Disadvantages
Fetal MRI	Uses a magnetic scanner to record detailed images of fetal organs and structures.	Provides the most detailed and accurate images available.	It is expensive. At present there is no evidence to suggest that it is harmful to the fetus.
Noninvasive prenatal testing (NIPT)	Cell-free fetal DNA are 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.

Source: Akolekar et al., 2015; Chan et al., 2013; Gregg et al., 2013; Odibo, 2015; Shahbazian et al., 2012; Shim et al., 2014; Theodora et al., 2016.

ENVIRONMENTAL INFLUENCES ON PRENATAL DEVELOPMENT

Prenatal development unfolds along a programmed path, a predictable pattern of change. However, environmental factors can interfere with the processes of prenatal development. A **teratogen** is an agent, such as a disease, drug, or other environmental factor, that disrupts prenatal development, increasing the risk of abnormalities and even death. 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

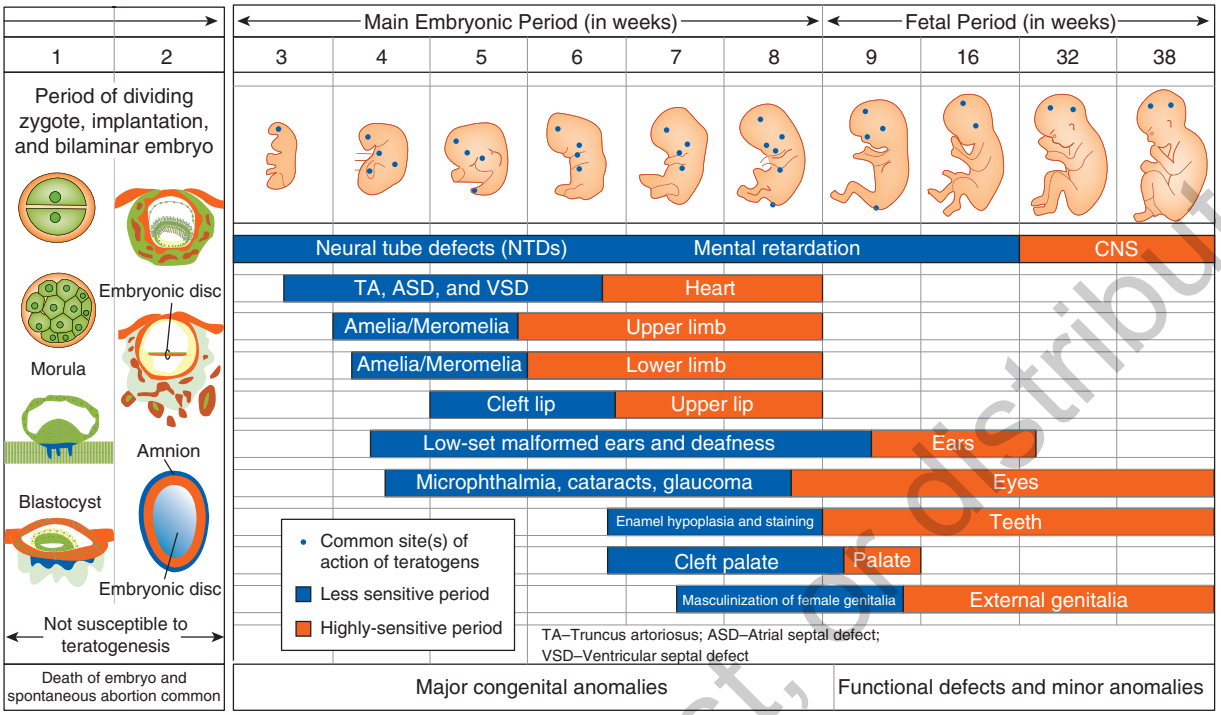
Several principles can account for the varied effects of exposure to teratogens on prenatal development.

Critical Periods

The extent to which exposure to a teratogen disrupts prenatal development depends on the stage of prenatal development when exposure occurs. During prenatal development, there are critical periods when the developing organism is more vulnerable to teratogens (Nelson & Gabard-Durnam, 2020). Exposure to teratogens during the germinal stage can interfere with cell division and prevent implantation. During this stage most women are unaware that they are pregnant and the effects often go unnoticed.

During the embryonic period the embryo is most sensitive to the harmful effects of teratogens (Webster et al., 2018). Structural abnormalities occur when the embryo is exposed to a teratogen while that part of the body is developing. Each organ of the body has a sensitive period in development during which it is most susceptible to damage from teratogens such as drugs, alcohol, and environmental contaminants (Figure 2.13). Once a body part is fully formed, it is less likely to be harmed by exposure to teratogens. Some body parts, like the brain, remain vulnerable throughout pregnancy.

FIGURE 2.13 ■ Critical Periods in Prenatal Development



Source: Adapted from Moore, K. L., Persaud, T. V. N., & Torchia, M. (2015). *Before we are born: Essentials of embryology and birth defects* (9th ed., Fig. 19.11). Elsevier.

Dose

The amount of exposure (i.e., dose level) to a teratogen influences its effects. Generally, the greater the dose and the longer the period of exposure, the more damage to development. Teratogens also differ in their strength. Some teratogens, like alcohol, display a powerful dose–response relationship so that larger doses, or heavier and more frequent drinking, result in greater damage (Bandoli et al., 2019).

Individual Differences

Individuals vary in their susceptibility to particular teratogens based on the genetic makeup of both the organism and mother. Teratogens increase the risk of abnormalities for all organisms, but responses may vary such that some organisms show severe problems, others more mild problems, and some may display normal development (Kaminen-Ahola, 2020). Dizygotic (fraternal) twins may show different effects in response to alcohol exposure in the womb, with one twin showing more adverse effects than the other (Astley Hemingway et al., 2019). The mother's genetic makeup and the prenatal environment may also increase or decrease the likelihood of teratogenic abnormalities.

Complicated Effects

Different teratogens can cause the same congenital abnormality, and a variety of abnormalities can result from the same teratogen. Also, some teratogenic effects may not be noticeable at birth but instead emerge later in life. (Charness et al., 2016). **Sleeper effects** refer to detrimental outcomes of exposure to teratogens and early risks that appear only later in development. Infants

born to women who consumed diethylstilbestrol (DES), a widely prescribed hormone between 1945 and 1970 to prevent miscarriages, were born healthy but as adults 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 birthweight (Conlon, 2017).

Types of Teratogens

Prenatal development can be influenced by many factors, such as exposure to prescription, nonprescription, and recreational drugs, and environmental factors, including chemicals, radiation, air pollution, and extremes of heat and humidity. In most cases women are unaware of their pregnancies until after the first few weeks of the embryonic stage are already past. Thus, no pregnancy can be entirely free of exposure to teratogens. Still, each year, about 97% of infants are born without congenital abnormalities (Centers for Disease Control and Prevention, 2020b; Mai et al., 2019). Next, we examine common teratogens.

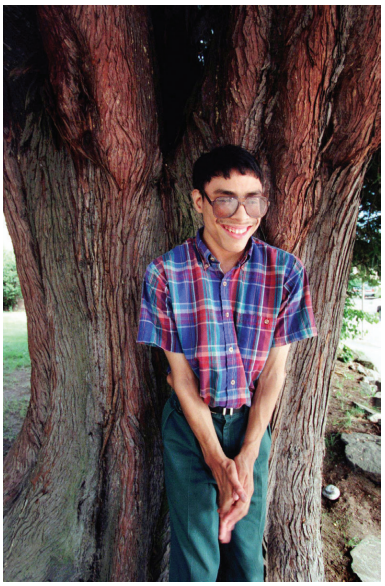
Prescription and Nonprescription Drugs

More than 90% of pregnant women take prescription or nonprescription medications (Servey & Chang, 2014; Stanley et al., 2019). Prescription drugs that can act as teratogens include antibiotics, certain hormones, antidepressants, anticonvulsants, and some acne drugs (Tsamantioti & Hashmi, 2020). In several cases, physicians have unwittingly prescribed drugs to ease pregnant women's discomfort that caused harm to the fetus. In the late 1950s and early 1960s, many pregnant women were prescribed a drug called thalidomide to prevent morning sickness. They later learned 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 (Fraga et al., 2016; Vargesson, 2019).

Nonprescription drugs, such as diet pills and cold medicine, can also cause harm, but research on over-the-counter (OTC) drugs lags far behind research on prescription drugs, and we know little about the teratogenic effect of many OTC drugs (Tsamantioti & Hashmi, 2020). Caffeine, found in coffee, tea, cola drinks, and chocolate, is the most common OTC drug consumed during pregnancy, yet its effects on prenatal development are disputed. Some research suggests that low doses of caffeine (200 milligrams or about one cup per day) may be safe (Modzelewska et al., 2019). Larger doses of caffeine are associated with an increased risk for miscarriage and low birthweight (Chen et al., 2014; Chen et al., 2016), leading some researchers to advise that women abstain from caffeine altogether (Qian et al., 2020).

Alcohol

About 10% to 20% of Canadian and U.S. women report consuming alcohol during pregnancy (Alshaarawy et al., 2016; Popova et al., 2017). Alcohol abuse during pregnancy is the leading cause of developmental disabilities (Webster et al., 2018). **Fetal alcohol spectrum disorders** refer to the continuum of effects of exposure to alcohol, which vary with the timing and amount of exposure (Hoyme et al., 2016). Fetal alcohol spectrum disorders are estimated to affect as many as 2% to 5% of younger schoolchildren in the United States and Western Europe (May et al., 2014, 2018). At the extreme end of the spectrum is **fetal alcohol syndrome (FAS)**, a cluster of congenital abnormalities appearing after heavy prenatal exposure to alcohol. 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 intellectual development, memory, visuospatial skills, motor coordination, and the combined



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.

Betty Udesen/KRT/Newscom

abilities to plan, focus attention, problem-solve, and use goal-directed behavior (Gupta et al., 2016; Loock et al., 2020; Wozniak et al., 2019). The effects of prenatal exposure to alcohol persist throughout childhood and adolescence and are associated with cognitive, learning, and behavioral problems from childhood and adolescence through adulthood (Dejong et al., 2019; Mamluk et al., 2017; Mattson et al., 2019).

Even moderate drinking is harmful because children may be born displaying some but not all of fetal alcohol syndrome problems, *fetal alcohol effects* (Hoyme et al., 2016). 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 (Flak et al., 2014; Kesmodel et al., 2019; Lundsberg et al., 2015). Even less than one drink per day has been associated with poor fetal growth, preterm delivery, and abnormal brain activity in newborns (Mamluk et al., 2017; Shuffrey et al., 2020). Sleeper effects may also occur with exposure to alcohol, as infants exposed prenatally to as little as an ounce of alcohol a day may display no obvious physical deformities at birth but later, as children, may demonstrate cognitive delays. Scientists have suggested that there is no safe level of drinking (Sarman, 2018; Shuffrey et al., 2020).

Cigarette Use and E-Cigarette Use

About 7% to 10%, and in some studies as many as 17%, of women report smoking cigarettes during pregnancy (Agrawal et al., 2019; Kondracki, 2019). Fetal deaths, premature births, and low birthweight are 1.5 to 2 times more frequent in mothers who are smokers than in those who do not smoke (Juárez & Merlo, 2013; Soneji & Beltrán-Sánchez, 2019). Infants exposed to smoke while in the womb are prone to congenital heart abnormalities, respiratory problems, and sudden infant death syndrome and, as children, show more behavior problems, have attention difficulties, and score lower on intelligence and achievement tests (Froggatt et al., 2020; He et al., 2020; Sutin et al., 2017). Moreover, maternal smoking during pregnancy shows epigenetic effects on offspring, influencing predispositions to illness and disease in childhood, adolescence, and even middle adulthood (Kaur et al., 2019; Nguyen et al., 2018). There is no safe level of smoking during pregnancy. Even babies born to light smokers (one to five cigarettes per day) show poorer fetal growth and higher rates of low birthweight than do babies born to nonsmokers (Berlin et al., 2017; Brand et al., 2019).

About 10% to 15% of women report using e-cigarettes during pregnancy and the prevalence is rising (Wagner et al., 2017; Whittington et al., 2018). E-cigarettes are commonly believed to the "safer" than traditional cigarettes, but there is no research to support this view (Holbrook, 2016). Animal research suggests that exposure to e-cigarette vapor prenatally is associated with increased risk for asthma, cognitive and neurological problems, and poor adjustment to stress (Church et al., 2020; Nguyen et al., 2018). Research with humans is sparse and just emerging, but it suggests that e-cigarettes have similar toxic effects on prenatal development as traditional cigarettes (Greene & Pisano, 2019; Whittington et al., 2018). Quitting cigarette smoking and e-cigarette use before or during pregnancy reduces the risk of adverse pregnancy outcomes (Soneji & Beltrán-Sánchez, 2019).

Marijuana

About 4% to 7% of pregnant women report using marijuana (Brown et al., 2017; Young-Wolff et al., 2019). The effects of marijuana, also referred to as cannabis, on prenatal development are

not well understood because there are few long-term studies of its effects and existing studies vary both in quality and in conclusions (El Marroun et al., 2018; Metz & Borgelt, 2018). The main active ingredient of marijuana, THC, readily crosses the placenta to affect the fetus, in lower doses than experienced by the mother (Alvarez et al., 2018). Marijuana use during early pregnancy negatively affects fetal length, growth and birthweight, preterm birth and is associated with a thinner cortex, the outer layer of the brain, in late childhood, suggesting that there are long-term neurological effects (El Marroun et al., 2016; Gunn et al., 2016). A growing body of research suggests that prenatal exposure to THC is associated with developmental delays as well as subtle long-term effects in cognition, including impairments in attention, memory, and executive function as well as impulsivity in children, adolescents, and young adults (Grant et al., 2018; Sharapova et al., 2018; Smith et al., 2016).

Cocaine

Prenatal exposure to cocaine is associated with low birthweight, impaired motor skills, difficulty managing arousal, and reduced brain volume at birth and in infancy (dos Santos et al., 2018; Grewen et al., 2014). Prenatal cocaine exposure has a small but lasting effect on attention and behavioral control, as well as language skills through early adolescence (Bazinet et al., 2016; Singer et al., 2015). In adolescence and emerging adulthood, prenatal exposure to cocaine is associated with emotional regulation, behavior problems, and substance use (Min et al., 2014; Richardson et al., 2019).

Opioids

Prenatal exposure to opioids, a class of drugs that include the illegal drugs heroin and synthetic opioids such as fentanyl, as well as pain relievers available legally by prescription, such as oxycodone, morphine, and others, poses serious risks to development. Newborns prenatally exposed to opioids may show addiction and withdrawal symptoms, such as tremors, irritability, abnormal crying, disturbed sleep, and impaired motor control (Conradt et al., 2019; Raffaeli et al., 2017). Prenatal exposure to opioids is associated with low birthweight, smaller head circumference, and altered brain development in newborns (Azuine et al., 2019; Towers et al., 2019). Children exposed to opioids prenatally tend to show difficulty with attention, managing arousal, learning, and inhibitory control (Bazinet et al., 2016; Levine & Woodward, 2018). Throughout childhood into adolescence, children exposed to opioids prenatally perform more poorly than their peers on tasks measuring intelligence and executive functioning (such as planning), show more emotional and behavioral problems, and, in adolescence, show reduced brain volume and smaller cortical surface area (Konijnenberg & Melinder, 2015; Nygaard et al., 2018; Yeoh et al., 2019).

Racial Disparities in Addressing Maternal Substance Use

Many U.S. states treat maternal substance use as fetal abuse and threaten pregnant women who use substances with involuntary treatment or protective custody (Atkins & Durrance, 2020; Seiler, 2016). About one half of U.S. states classify controlled substance use during pregnancy as child abuse and one half of U.S. states require that substance use by pregnant mothers be reported to child protective services, which may lead to removing the newborn from parental custody or even terminating parental rights altogether (Guttmacher Institute, 2020). In some cases, these consequences have been extended to include alcohol abuse and dependence (Paltrow & Flavin, 2013; Seiler, 2016). As of 2021, 40 states had laws related to reporting of alcohol consumption during pregnancy (Alcohol Policy Information System, 2021).

Policies criminalizing maternal substance use discriminate against women of color and those in low SES brackets because low-income Black and Hispanic women are disproportionately tested and reported to child protective services for substance use (Paltrow & Flavin, 2013). In one California county with universal screening policies, Black and white pregnant women showed similar rates of drug and alcohol use, but Black women were 4 times more likely than white women to be reported to child protective services after delivery (Roberts & Nuru-Jeter, 2012). Other recent research confirms that Black and Native American mothers are more likely to be reported for alcohol use and marijuana than are white mothers (Hoerr et al., 2018; Rebbe et al., 2019a, 2019b)

Criminal sanctions for maternal drug can discourage women from seeking prenatal and postnatal care and undermine the physician–patient relationship (American College of Obstetricians and Gynecologists, 2011; American Medical Association, 2014). Such policies can cause women to mistrust medical professionals, which ultimately harms their care if they become reluctant to seek medical care for themselves and their children. One recent study of state policies from 2000 to 2012 found that punitive prenatal substance use policies were ineffective because they were not linked with a reduction in maternal substance abuse exposure at birth (Atkins & Durrance, 2020). Instead, these policies may deter women from seeking substance use treatment during pregnancy. In contrast, women who live in states that adopt multiple policies, including treatment and support, are more likely to seek treatment (Kozhimannil et al., 2019). Fetal outcomes are supported by substance abuse treatment that rewards abstention, invests in family and community supports, and promotes contact with health care and social support services (Bada et al., 2012; Hui et al., 2017).

Context–Teratogens Interactions

Our discussion of teratogens thus far has examined the effects of each teratogen independently, which is somewhat misleading because infants are often exposed to multiple teratogens. For example, most infants exposed to opioids or cocaine were also exposed to other substances with teratogenic effects, including tobacco, alcohol, and marijuana, making it difficult to isolate the effect of each drug on prenatal development. Mothers who use two substances, such as opioids and marijuana, are more likely to give birth to premature and low birthweight infants than are those who use one, such as opioids alone (Stein et al., 2020).

In addition, we must be cautious in interpreting findings about illicit drug use and the effects on development because the effects of prenatal exposure to drugs are influenced by parenting and other postnatal (after birth) factors including poverty, inconsistent parenting, and stress (S. J. Lee et al., 2020; Smith et al., 2016). Parents who abuse substances are more likely 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 (Hatzis et al., 2017).

Quality care can lessen the long-term impact of prenatal exposure to substances (Beach et al., 2010;Brodie et al., 2019; Calhoun et al., 2015). When medical and environmental postnatal factors are considered, developmental differences in substance-exposed infants are reduced and often disappear (Behnke & Smith, 2013). 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.

Thinking in Context

1. Consider the influence of teratogens from the perspective of Bronfenbrenner's bioecological systems theory (Chapter 1). Identify examples of teratogens at each bioecological level: microsystem, macrosystem, exosystem, and macrosystem. How might this model be used to help promote healthy prenatal development?
2. Suppose that you plan to study the presence and effects of teratogens on prenatal development. Choose a teratogen that you believe is most relevant to prenatal health.
   a. How might you measure the fetus or embryo's exposure to the teratogen? What effects would you study?
   b. To what extent are other teratogens likely to be present? How might this complicate your results?
   c. How will you obtain participants, pregnant women? How might you ensure that your participants are diverse in terms of race, ethnicity, and SES? Are there other relevant variables on which women might differ?
   d. In what ways might interactions among race, ethnicity, and SES influence your results, if at all? Why or why not?

THE PRENATAL ENVIRONMENT AND PRENATAL CARE

Parental characteristics (of both biological mother and biological father) influence the prenatal environment. A pregnant woman's characteristics, such as her diet, emotional well-being, and age influence prenatal outcomes. The biological father, or sperm donor, passes on characteristics and risks genetically (as discussed earlier in this chapter) and epigenetically, through behaviors that affect sperm.

Maternal Nutrition

To sustain a healthy pregnancy, most women need to consume 2,200 to 2,900 calories per day (and gain about 25 to 30 pounds in total) (Kaiser et al., 2008). Fetal malnutrition is associated with poor growth before and after birth as well as effects that can last into adulthood, including vision, motor, and speech disabilities; cognitive impairment; and increased risk of heart disease, stroke, and diabetes in adulthood (Han & Hong, 2019; Kim et al., 2017). Infants who are malnourished can overcome some of the negative effects if they are raised in enriched environments with adequate food and health care. Still, most children who are malnourished before birth remain malnourished; few are raised in enriched environments after birth.

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

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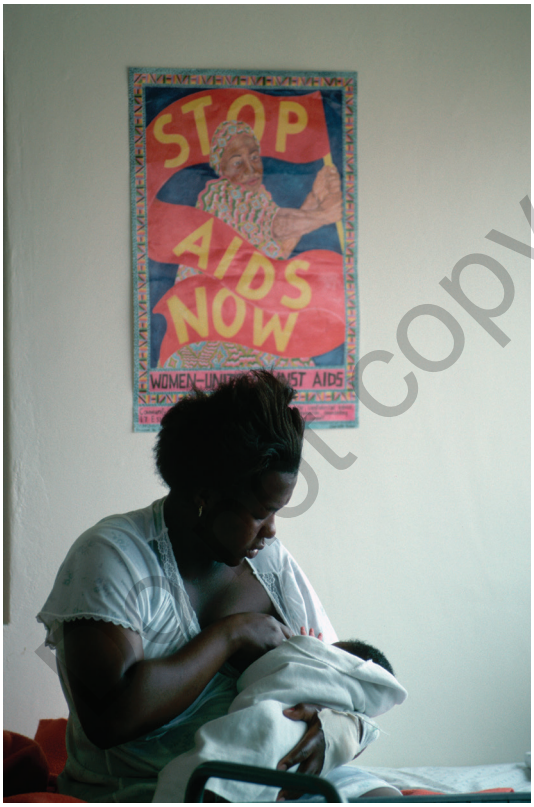
Prenatal development relies on obtaining adequate nutrients. For instance, inadequate consumption of folic acid (a B vitamin) very early in pregnancy can result in the formation of neural tube abnormalities stemming from the failure of the neural tube to close. **Spina bifida** occurs when the lower part of the neural tube fails to close and spinal nerves begin to grow outside of the vertebrae, often resulting in paralysis (Avagliano et al., 2019). Spina bifida is often accompanied by malformations in brain development and impaired cognitive development (Donnan et al., 2017). Surgery must be performed before or shortly after birth, but lost capacities cannot be restored (Adzick, 2013; Dewan & Wellons, 2019).

Another neural tube abnormality, **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 (Avagliano et al., 2019). Neural tube abnormalities can be prevented by consuming .4 to .8 milligrams of folic acid. Many foods are fortified with folic acid. A dietary supplement is safe and ensures that prenatal needs are met, but the majority of pregnant women consume less than the recommended dose (Bibbins-Domingo et al., 2017; Tinker et al., 2010).

Maternal Illness

Depending on the type and time of occurrence, an illness experienced during pregnancy can have grave consequences for the developing fetus. Rubella (German measles) prior to the 11th week of pregnancy can cause a variety of congenital abnormalities, including blindness, deafness, heart abnormalities, and brain damage, but after the first trimester, adverse consequences become less likely (Bouthry et al., 2014; Singh, 2020). Chicken pox can produce congenital abnormalities affecting the arms, legs, eyes, and brain; mumps can increase the risk of miscarriage (Mehta, 2016; Webster et al., 2018). Children born to women infected with the mosquito-borne Zika virus are at risk for *congenital Zika syndrome*, which includes a pattern of abnormalities, such as 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 (D. S. Moore, 2017; Veiga et al., 2021).



HIV can be transmitted from mother to infant through breastfeeding.

David Turnley/Contributor/Getty Images

HIV, the virus that causes *acquired immune deficiency syndrome (AIDS)*,a disease affecting the immune system, can be transmitted prenatally through the placenta, by exposure to body fluids, and breastfeeding. Infants and children with HIV are at high risk for a range of illnesses and health conditions, including heart, gastrointestinal, and lung problems; growth stunting; problems in brain development, which contribute to cognitive and motor impairment; and delays in reaching developmental milestones (McHenry et al., 2018; Wedderburn et al., 2019).

The use of cesarean delivery and prescribing anti-HIV drugs to the mother during the second and third trimesters of pregnancy and to the infant for the first 6 weeks of life have reduced the rate of mother-to-child transmission of HIV to about 1% in the United States and Europe (Blanche, 2020; Selph et al., 2019). Over two thirds of the HIV-infected children born from 2002 to 2013 were to Black or African American mothers (63%) and about 18% to Hispanic or Latina

mothers (Taylor et al., 2017). A combination of socioeconomic factors influences these health disparities, such as lack of health insurance, limited health literacy, poverty, and associated sense of powerlessness, which may prevent women from seeking assistance. HIV medications and treatment are expensive and an HIV diagnosis is often stigmatizing and may alienate individuals from their communities. Aggressive treatment may further reduce the transmission of HIV to newborns, and research suggests that it may even induce remission (Blanche, 2020; Rainwater-Lovett et al., 2015). However, women of color and those in poverty are less likely to receive HIV treatment.

Maternal Emotional Well-Being

Exposure to stress, such as by living in an unsafe environment, experiencing traumatic life events, and experiencing racism, stigma, and discrimination, poses risks for prenatal development, including low birthweight and premature birth (Lima et al., 2018; Schetter & Tanner, 2012). 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 and greater release of stress hormones in response to everyday discomfort, such as bathing (McGowan & Matthews, 2018; Nazzari et al., 2019). As a result, newborns exposed to prenatal stress tend to be more irritable and may have difficulties in sleep, digestion, and self-regulation. In childhood, prenatal stress is associated with sleep problems, emotional difficulties, behavior problems, and an increased risk for autism and attention deficit/hyperactivity disorder (Hentges et al., 2019; MacKinnon et al., 2018; Manzari et al., 2019; Simcock et al., 2019).

Prenatal stress may also have epigenetic effects on development, influencing stress responses throughout the lifespan, especially in marginalized groups (Conradt et al., 2020; DeSocio, 2018). Animal research has suggested that the epigenetic effects of prenatal exposure to stress may be transmitted across generations, potentially affecting multiple cohorts of individuals (Badihian et al., 2020).

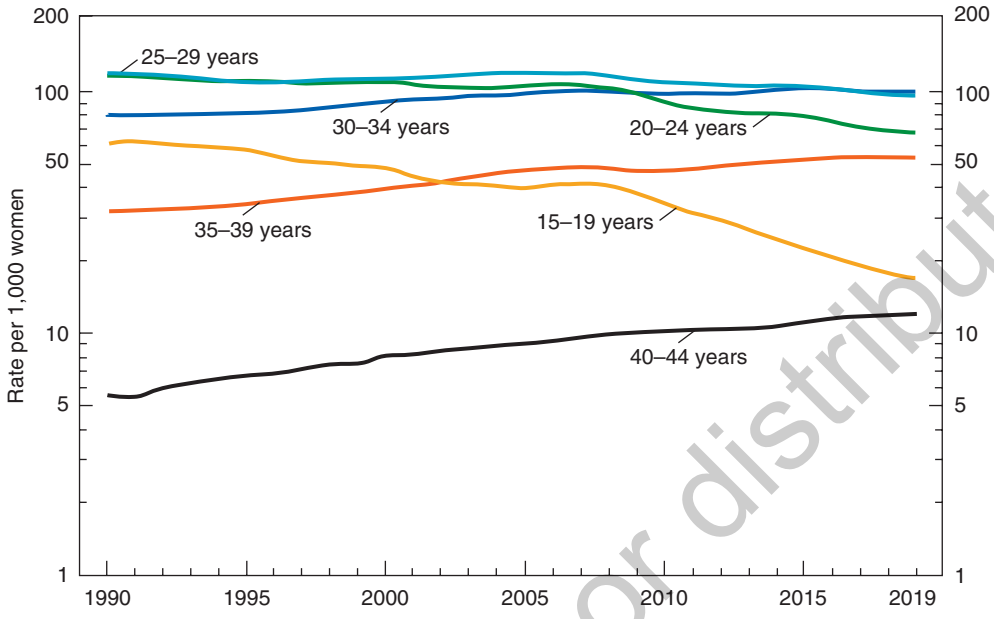
Maternal Age

U.S. women are becoming pregnant at later ages than ever before. Since 1990, the birth rate has increased for women ages 35 to 39 and 40 to 44 and decreased slightly for women in their 20s (Hamilton et al., 2017) (Figure 2.14). Does maternal age matter? The risk of birth complications increases in the late 30s, especially after age 40. Women who give birth when they are older than age 40 are at greater risk for pregnancy and birth complications, including hypertension, gestational diabetes, preterm birth, and miscarriage than younger women (Londero et al., 2019; Magnus et al., 2019; Marozio et al., 2019). These involve increased risks to the newborn, including low birthweight, preterm birth, respiratory problems, and related conditions requiring intensive neonatal care (Frederiksen et al., 2018; Grotegut et al., 2014; Kenny et al., 2013; Khalil et al., 2013).

The risk of having a child with Down syndrome also increases sharply with maternal age, especially after age 40 (Diamandopoulos & Green, 2018; Hazlett et al., 2011) (Figure 2.15).

Although risks for complications rise linearly with each year, it is important to realize that the majority of women over age 35 give birth to healthy infants (Yaniv et al., 2011). Differences in context and behavior may compensate for some of the risks of advanced maternal age. For example, longer use of oral contraceptives prior to conception is associated with a lower risk of giving birth to a child with Down syndrome (Horányi et al., 2017; Nagy et al., 2013).

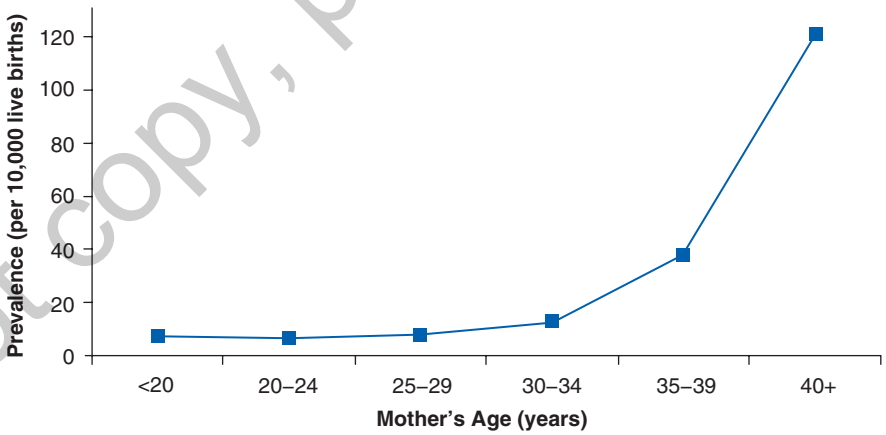
FIGURE 2.14 ■ Birth Rates by Age of Mother, United States 2019



Source: Martin, J. A., Hamilton, B. E., Osterman, M. J. K., & Driscoll, A. K. (2021). Births: Final data for 2019. *National Vital Statistics Reports, 70*(2). https://doi.org/10.15620/CDC:100472

FIGURE 2.15 ■ Maternal Age and Down Syndrome

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



Centers for Disease Control and Prevention. (2019). *Data and statistics on Down syndrome*. https://www.cdc.gov/ncbddd/birthdefects/downsyndrome/data.html

Paternal Characteristics and Prenatal Development

It was once thought that biological fathers had no influence on prenatal development and therefore researchers neglected to study their role. Most obviously, fathers influence the home context. Secondhand smoke from fathers is harmful to the developing organism (Braun et al., 2020). Fathers' interactions with pregnant mothers can increase maternal stress, with potential negative

implications for prenatal development, but fathers can also be important sources of social support, aiding mothers (Glover & Capron, 2017). We know less about how fathers' health, behaviors, and contextual factors act as biological influences on prenatal development

Advanced paternal age is associated with an increased risk of congenital abnormalities, chromosomal abnormalities, and developmental disorders such as Down syndrome, and autism spectrum disorder (Brandt et al., 2019; Day et al., 2016; Herati et al., 2017). Advanced age (over 40) is associated with damage to sperm and DNA (Rosiak-Gill et al., 2019). Alcohol and substance use and exposure to toxins such as lead can impair sperm production and quality (Borges et al., 2018; Estill & Krawetz, 2016). Smoking is associated with DNA damage and mutations in sperm (Beal et al., 2017; Esakky & Moley, 2016).

In addition to DNA, fathers (and mothers) pass on epigenetic markers that can influence their offspring's health throughout life. In one study, men whose fathers smoked when they were conceived had a 50% lower sperm count than the men of nonsmoking fathers (Axelsson et al., 2018). Epigenetic marks can be passed potentially from generation to generation (Bošković & Rando, 2018). But it is important to remember that the epigenetic marks we are born with are not set in stone. Some epigenetic marks can be changed after birth through experiences, health care, and behaviors, such as diet and exercise (Champagne, 2018).

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. Prenatal care visits typically include a physical exam, weight check, and diagnostic procedures to assess the fetus's health. These visits also provide women the opportunity to ask questions and for the service provider to provide health care information and advice about nutrition, prenatal care, and preparing for birth. Unfortunately, not all women obtain early prenatal care.

Barriers to Prenatal Care

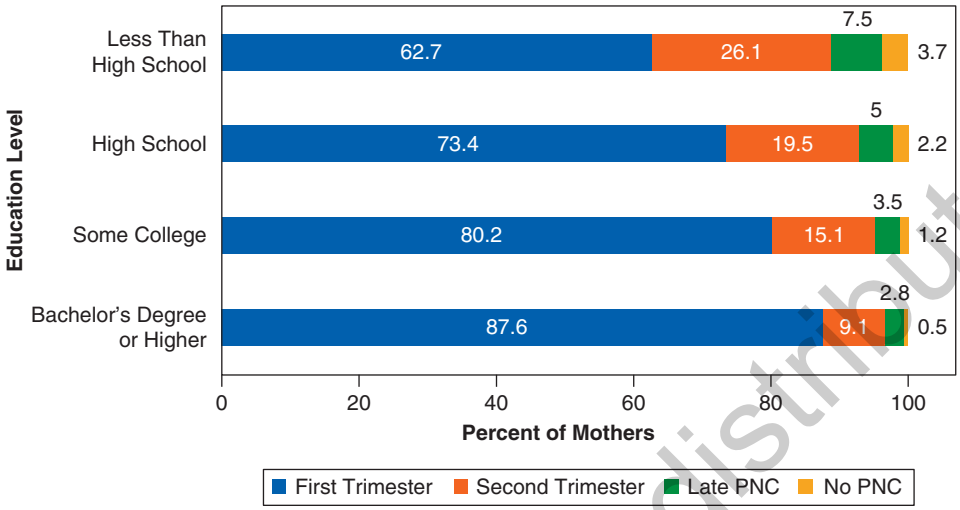
About one quarter of pregnant women in the United States do not obtain prenatal care until after the first trimester; 6% obtain prenatal care at the end of pregnancy or not at all (U.S. Department of Health and Human Services, 2014). Inadequate prenatal care is a risk factor for low-birthweight and preterm births as well as infant mortality during the first year (Partridge et al., 2012; Xaverius et al., 2016). In addition, use of prenatal care predicts the use of pediatric care, and thereby health and development, throughout childhood (Deaton et al., 2017).

Why do women delay or avoid seeking prenatal care? A common reason is the lack of health insurance (Baer et al., 2019). Government-sponsored health care is available for the poorest mothers, but many low-income mothers do not qualify for care or lack information on how to take advantage of care that may be available. Other common barriers to obtaining prenatal care include lacking transportation, not being able to take time off from work, and lacking child care (Daniels et al., 2006; Heaman et al., 2015; Mazul et al., 2017). Black and Latina women report nearly twice as many barriers to accessing care than white women (Fryer et al., 2021).

Race, Ethnicity, and Prenatal Care

There are significant ethnic and socioeconomic disparities in prenatal care. As shown in Figure 2.16, prenatal care is closely linked with maternal education (Blakeney et al., 2019). About 86% of women with a college degree obtain first-trimester care, compared with less than two thirds of women with less than a high school diploma (U.S. Department of Health and Human Services, 2014).

FIGURE 2.16 ■ Timing of Prenatal Care (PNC) by Age and Education, 2016



Source: Osterman, M. J. K., & Martin, J. A. (2018, May 30). Timing and adequacy of prenatal care in the United States. *National Vital Statistics Reports*, 67(3).

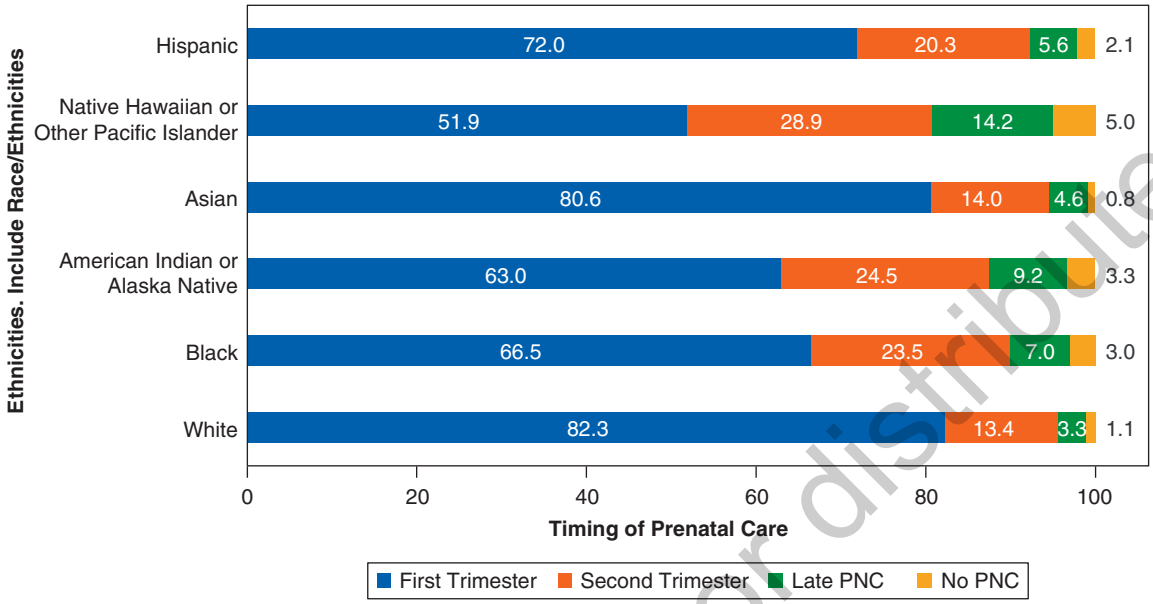
Women of color are disproportionately less likely to receive prenatal care during the first trimester and are more likely to receive care beginning in the third trimester or no care (Blakeney et al., 2019). In 2018, Native Hawaiian and Native American women were least likely to obtain prenatal care during the first trimester, followed by Black, Hispanic, and, Asian American and white American women (Hamilton et al., 2019) (Figure 2.17). Ethnic differences are thought to be largely influenced by socioeconomic factors. The ethnic groups least likely to seek early prenatal care are also the most economically disadvantaged members of society and are most likely to live in communities with fewer health resources, including access to physicians and hospitals, sources of health information, and nutrition and other resources.

Prenatal care predicts better birth outcomes, but cultural factors protect some women and infants from the negative consequences of inadequate prenatal care. Known as the *Latina paradox*, Latina mothers, despite low rates of prenatal care, tend to experience low birthweight and mortality rates below national averages. These favorable birth outcomes are striking because of the strong and consistent association between SES and birth outcomes. Latinx are among the most socioeconomically disadvantaged ethnic populations in the United States (McGlade et al., 2004; Ruiz et al., 2016).

Several factors may account for the Latina paradox, including strong cultural support for maternity, healthy traditional dietary practices, and the norm of selfless devotion to the maternal role (known as *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 in which women help other women in the community, and warm interpersonal relationships, known as *personalismo*, are highly valued (Fracasso & Busch-Rossnagel, 1992; McGlade et al., 2004).

These cultural factors are thought to underlie the positive birth outcomes seen in Latina women, yet they appear to erode as Latina women acculturate to American society. The birth advantage declines in subsequent American-born generations. Some researchers have called

FIGURE 2.17 ■ Timing of Prenatal Care by Race and Ethnicity, 2016



Source: Osterman, M. J. K., & Martin, J. A. (2018, May 30). Timing and adequacy of prenatal care in the United States. *National Vital Statistics Reports*, 67(3).

the existence of the Latina paradox into question, as some samples have illustrated that cultural supports cannot easily ameliorate the negative effects of socioeconomic disadvantage (Hoggatt et al., 2012; Sanchez-Vaznaugh et al., 2016). Other research suggests that the Latina paradox is more complex. One recent study of Puerto Rican women found that women with bicultural acculturation, who identify with both Latina and continental U.S. cultures experience lower stress levels than those with low acculturation (Chasan-Taber et al., 2020). Women's sense of identity may play a role in their perceived stress and perhaps, ultimately, the health of their pregnancy.

The mixed findings regarding the Latina paradox may also be influenced by complex intersecting social factors, such as geography and politics. Latina women who live on the U.S.–Mexico border are much more likely to have a cesarean birth than those who do not live on the border. This strip of land spans 2,000 miles and four states (Morris et al., 2018). Hot political debates center around this strip, including debates about immigration, poverty, crime, and whether to construct a physical barrier, a wall, to distinguish each country. The intersecting social disadvantages for Latina women who live on the border may pose multiple threats to their health and the health of their fetus.

Thinking in Context

1. To what degree do the influence of maternal and paternal characteristics illustrate nature (biology) or nurture (environment)? Explain your reasoning.
2. What are some examples of barriers to receiving prenatal care? In what ways do factors such as race, ethnicity, SES, and culture influence whether women receive prenatal care? What environmental factors might contribute to these differences?

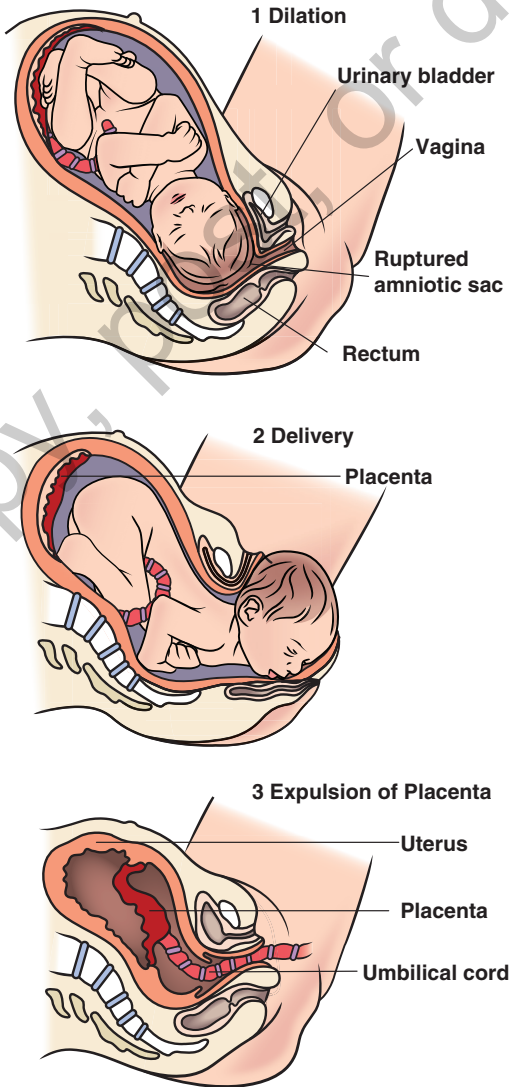
CHILDBIRTH

At about 40 weeks of pregnancy, or 38 weeks after conception, childbirth, also known as **labor**, begins.

Labor

Labor progresses in three stages. The first stage of labor, dilation, is the longest. It typically lasts 8 to 14 hours for a woman having her first child; the average is 3 to 8 hours for later-born children. Labor begins when the mother experiences regular uterine contractions spaced at 10- to 15-minute intervals. Initial contractions may feel like a backache or menstrual cramps or may be extremely sharp. The amniotic sac, a membrane containing the fetus surrounded by fluid, may rupture at any time during this stage, often referred to as the "water breaking." The contractions, which gradually become stronger and closer together, cause the cervix to dilate so that the fetus's head can pass through, as shown in Figure 2.18.

FIGURE 2.18 ■ Stages of Labor



The second stage of labor, delivery, begins when the cervix is fully dilated to 10 cm 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. During this stage, the mother typically feels an urge to push or bear down with each contraction to assist the birth process. Delivery can take from 30 minutes to an hour and a half.

In the third stage of labor, the placenta separates from the uterine wall and is expelled by uterine contractions. This typically happens about 5 to 15 minutes after the baby has emerged. The process can take up to a half hour.

Medication During Delivery

Medication, most commonly *anesthetics*,painkillers that block body sensations, are administered in most births in the United States (Declercq et al., 2014). Anesthetics are frequently in the form of an *epidural,* in which the drug is administered to a small space between the vertebrae of the lower spine, numbing the woman's lower body. Epidurals are associated with a longer delivery as they weaken uterine contractions and may increase the risk of a cesarean section, as discussed next (Gabbe et al., 2016; Herrera-Gómez et al., 2017). An analysis of nearly 15,500 deliveries suggested that newborns exposed to epidural anesthesia did not differ from those exposed to no anesthesia (Wang et al., 2018). The American College of Obstetricians and Gynecologists (2017) has concluded that the proper administration of medication poses few risks to the newborn and that pain medication should be available to all women.

Cesarean Delivery

Sometimes a vaginal birth is not possible because of concerns for the health or safety of the mother or fetus. At birth the fetus is positioned with its head closest to the cervix. A fetus in **breech position** is positioned in reverse, with its feet or bottom closest to the cervix. During a breech birth a fetus may become stuck in the birth canal and the oxygen supply through the umbilical cord may be cut off, causing irreversible damage and even death. Sometimes the obstetrician can turn the baby so that it is head-first. In other cases the fetus is removed from the uterus through the abdomen in a surgical procedure known as a **cesarean delivery**.

About 32% of U.S. births were by cesarean delivery in 2018 (Hamilton et al., 2019; J. A. Martin et al., 2018) Cesarean deliveries are also performed when labor progresses too slowly, the head is too large to pass through the pelvis, or the fetus or mother is in danger (Jha et al., 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 are similar for infants delivered vaginally and by cesarean (Durik et al., 2000).

Natural Childbirth

Natural childbirth is an approach to birth that reduces pain through the use of breathing and relaxation exercises. Natural childbirth methods emphasize preparation by pregnant women and their partners about childbirth, helping them reduce their fear, and teaching them pain management techniques. The most widely known natural childbirth method—the Lamaze method—was created by a French obstetrician, Ferdinand Lamaze (1956). The Lamaze method emphasizes reducing women's fear and anxiety about labor though education about their bodies, including detailed information about anatomy, pregnancy, and childbirth. When women know

what to expect and learn a breathing technique to help them relax, they are better able to manage the pain of childbirth. The Lamaze method relies on the spouse or partner as coach, providing physical and emotional support.

In addition to the expectant mother's partner, a doula can be an important source of support. A **doula** is a caregiver who gives support to an expectant mother and her partner throughout the birth process (Kang, 2014). Doulas provide education about pregnancy, delivery, and pain management practices and support the woman in creating a birth plan. The doula is present during birth, whether at a hospital or other setting, and helps the woman carry out her birth plans. The presence of a doula is associated with less pain medication, fewer cesarean deliveries, and higher satisfaction rates in new mothers (Gabbe et al., 2016; Kozhimannil et al., 2016).

Home Birth

Common in nonindustrialized nations, home birth is rare, comprising 1.5% of all births in 2016 in the United States (MacDorman & Declercq, 2016). The remaining 98% of births occur in hospitals. Most home births are managed by a **midwife**, a health care professional, and usually a nurse, specializing in childbirth. Midwives provide health care throughout pregnancy and supervise home births. One review of 50 studies found that the use of midwives, whether as part of a home birthing plan or as part of a plan to birth in a hospital setting, is associated with reduced neonatal mortality, reduced preterm birth, fewer interventions, and more efficient use of medical resources (Renfrew et al., 2014).

Is a home birth safe? A healthy woman, who has received prenatal care and is not carrying twins, is unlikely to encounter problems requiring intervention—and may be a good candidate for a home birth (Wilbur et al., 2015). Although unpredictable events can occur and immediate access to medical facilities can improve outcomes, studies from Europe indicate that home birth is not associated with greater risk of perinatal mortality. Home birth is far more common in many European countries than in the United States (20% in the Netherlands, 8% in the United Kingdom, and about 1% in the United States) (Brocklehurst et al., 2011; de Jonge et al., 2015). The few U.S. studies examining planned home birth compared with hospital birth have found no difference in neonatal deaths or Apgar scores (the Apgar is described later in this section), and women who have a planned home birth report high satisfaction rates (Jouhki et al., 2017; Zielinski et al., 2015).

Cultural Childbirth Practices

Societies vary in birth customs, 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 within designated hospital visiting hours; the newborn usually rooms with the mother all or part of the day.

Cultures vary in birthing practices. 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). A few days before and until about 1 month after the birth, the mother-in-law brings and feeds the mother ritual foods of broth, marsala, and fresh cheeses. Just after birth, the midwife brings the mother's immediate and extended family to the mother's room and they take turns congratulating the mother and baby, kissing them. During labor and afterward, the mother is and visited by many of her friends and relatives to recognize the mother's contribution to the community.

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, husbands, children, and other family members attend the birth, which occurs in the home with the aid of a midwife and female relatives. Balinese women know what to expect in giving birth to their first child because they have been present at many births (Diener, 2000). Babies are believed to be reincarnated souls of ancestors and are immediately integrated into the family and community. Many kin are present to support the mother and baby since the child is viewed as related to many more people than its parents.

An Uzbekistan midwife prepares to deliver a baby by first listening to its heartbeat.

Peter Turnley/Contributor/Getty Images

Among the Maya of the Yucatan region of Mexico, there are few changes in the expectant mother's surroundings during labor. The Mayan woman lies in the same hammock in which she sleeps each night. The father-to-be is present during labor and birth to assist and to witness the suffering that accompanies labor. The pregnant woman's mother is present, often in the company of sisters, sisters-in-law, mothers-in-law, godmothers, and sometimes neighbors and close friends. The mother and child remain inside the house for 1 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).

Newborn Health Screening

After birth, newborns are routinely screened with the **Apgar scale**, which provides a quick 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 5 minutes after birth; this timing ensures that hospital staff monitors the newborn over several minutes. A low APGAR score at both time points is associated with an increased risk of neonatal death (Chen et al., 2014). Over 98% of all newborns in the United States achieve a 5-minute score of 7 to 10, indicating good health (J. A. Martin et al., 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: Adapted from Apgar, V. (1953). A proposal for a new method of evaluation in the newborn infant. *Current Research in Anesthesia and Analgesia*, *32*, 260–267.

Low-Birthweight and Preterm Infants

About 8% of infants in the United States each year are born with low birthweight (J. A. Martin et al., 2018). Infants are classified as **low birthweight** when they weigh less than 2,500 grams (5.5 pounds) at birth; *very low birthweight* refers to a weight less than 1,500 grams (3.5 pounds), and *extremely low birthweight* refers to a weight less than 750 grams (1 lb., 10 oz.). Low-birthweight (LBW) infants may be **preterm,**premature(born before their due date), or **small for date**, full term but have experienced slow growth and are smaller than expected for their gestational age. Infants who are born with low birthweight are at risk for a variety of developmental difficulties. Indeed, their very survival is far from certain. Low birthweight is the second leading cause of infant mortality (Murphy et al., 2018) (Mathews & MacDorman, 2013).

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

Chip Somodevilla/Staff/Getty Images

Characteristics of LBW Infants

At birth, many LBW infants experience difficulty breathing and are likely to suffer from respiratory distress syndrome, in which they breathe irregularly and at times may stop breathing (Charles et al., 2018). 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.

The developmental and health problems that LBW infants experience correspond closely to the infant's birthweight, with extremely low-birthweight infants suffering the greatest problems (Hutchinson et al., 2013). LBW infants are at higher

risk for poor growth, cerebral palsy, seizure disorders, neurological difficulties, respiratory problems, and illness (Charles et al., 2018; Durkin et al., 2016; J. E. Miller et al., 2016). In childhood they often experience difficulty in self-regulation, inattention, hyperactivity, and cognitive and social problems that may persist into adulthood (Eryigit-Madzwamuse et al., 2015; Franz et al., 2018; Jaekel et al., 2018; Mathewson et al., 2017).

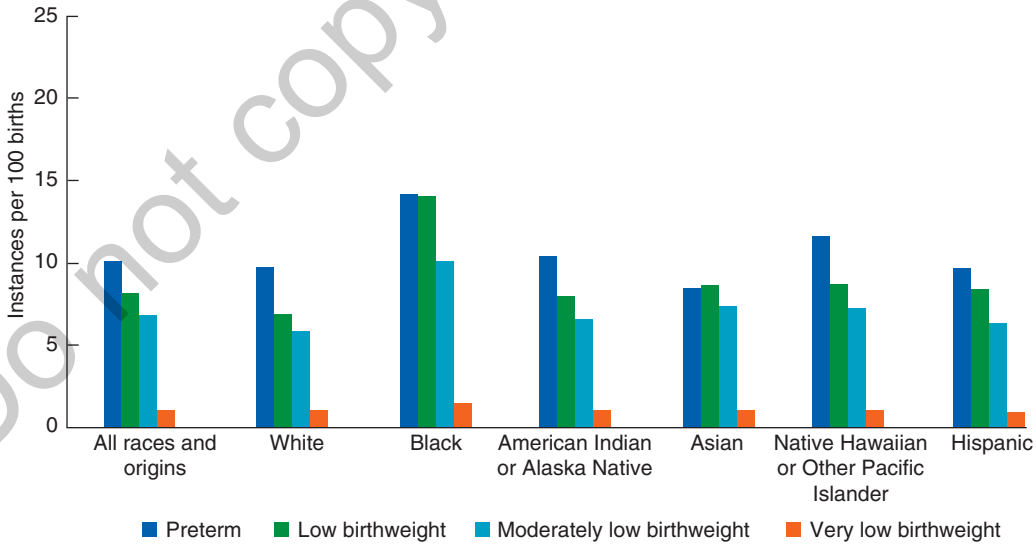
Race, SES, and LBW

Socioeconomic disadvantage, race, and LBW are complexly interwoven in the United States (Figure 2.19). In 2016 non-Hispanic Black infants were more than twice as likely to be born low birthweight (11%) than non-Hispanic white and Hispanic infants (5% and 6%, respectively) (Womack et al., 2018). SES plays a role in these differences, but it is not the whole story. In one study, LBW rates were higher in non-Hispanic Black mothers than non-Hispanic white mothers; the racial difference declined (but did not disappear) when the researchers took into account financial and relationship stresses, suggesting a role for SES in racial differences but also other factors (Almeida et al., 2018). In another study of over 10,000 Californian women, the most economically disadvantaged Black and white women showed similar LBW rates, but increases in income were more strongly associated with improvement in LBW rates among white than Black women (Braveman et al., 2015). As SES advantage increased for both white and Black women, the racial disparity in LBW outcomes grew. Racial differences in LBW are a function not only of income but of other factors such as the experience of racism and discrimination (Ncube et al., 2016; Ramraj et al., 2020).

Caring for LBW Infants

Sensitive caregiving helps LBW infants adjust and thrive. One popular and effective intervention known as **kangaroo care** involves skin-to-skin contact between infant and caregiver (Charpak et al., 2005). The infant is placed upright against the caregiver's chest, under the shirt. As the

FIGURE 2.19 ■ Gestational Age and Birthweight: United States, 2018



Source: Hamilton, B. E., Martin, J. A., Osterman, M. J. K., & Rossen, L. M. (2019). Births: Provisional Data for 2018. In *NVDD vital statistics rapid release*. https://www.cdc.gov/nchs/products/index.htm.

caregiver 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. 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., 2016; Jefferies, 2012; Sharma et al., 2019).

Interventions to promote the development of low-birthweight children help parents learn coping strategies for interacting with their infants and managing parenting stress (Chang et al., 2015; Lau & Morse, 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-birthweight infants tend to have good long-term outcomes, often catching up to their peers (Jaekel et al., 2015; Jones et al., 2009; J. L. Lynch & Gibbs, 2017). One study of low-birthweight 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, but those who experienced below-average levels of sensitive parenting showed lasting deficits (Camerota et al., 2015).

Thinking in Context

1. Why do cultures vary in birth practices? What kinds of factors might underlie the differences we observe? Relatedly, how might cultural and contextual factors influence the rates of home birth, natural birth, cesarean birth?
2. Ask adults of different generations, perhaps a parent or an aunt and a grandparent or family friend, about their birth experiences. How do these recollections compare with current birthing practices?
3. A basic tenet of development is that individuals are active in their development, influencing the world around them (Chapter 1). Consider low-birthweight infants: How might their characteristics and abilities influence their caregivers? Why is caring for LBW infants challenging?
4. How might contextual factors influence parents' responses to their low-birthweight infants? What supports from the family, community, and broader society can aid parents in helping their low-birthweight infants adapt and develop healthily?

APPLY YOUR KNOWLEDGE

Strapped in and buckled in the rear seat of her mother's bicycle, 1-year-old Jenna patted her helmet as her mother zoomed along the bike path to the beach. She giggled and kicked her legs as her mother whooshed her through the water. As a child, Jenna loved to be outside and especially in the water. Jenna practiced swimming at the local YMCA nearly every day and became quite skilled. Jenna's proud mother encouraged her daughter's athleticism by enrolling her in swim classes. As a teenager, Jenna decided that if she were going to become an exceptional swimmer, she would have to go to a summer swimming camp. She researched camps and asked her mother if she could attend. Jenna further honed her skills as a swimmer and won a college scholarship for swimming.

Many years later, Jenna was surprised to learn that she had a twin sister, Tasha. Separated at birth, Jenna and Tasha became aware of each other in their early 40s. Jenna was stunned yet couldn't wait to meet her twin sister. Upon meeting, Jenna and Tasha were surprised to find that

they were not exactly the same. Whereas Jenna was athletic and lithe, Tasha was more sedentary and substantially heavier than Jenna. Unlike Jenna, Tasha grew up in a home far from the beach and with little access to outdoor activities. Instead Tasha's interest was writing. As a child, she'd write stories and share them with others. She sought out opportunities to write and chose a college with an exceptional writing program. Both Jenna and Tasha excelled in college, as they did throughout their education, and earned nearly identical scores on the SAT.

Jenna and Tasha look very similar. Even the most casual observer could easily tell that they are sisters as both have blond hair, blue eyes, and a similar facial structure. Tasha's skin, however, is more fair and unlined. Jenna's face is sprinkled with freckles and darker spots formed after many days spent swimming outside. Both Jenna and Tasha are allergic to peanuts, and they both take medication for high blood pressure. The more that Jenna and Tasha get to know each other, the more similarities they find.

1. Considering Jenna and Tasha, provide examples of three types of gene–environment correlations: passive, evocative, and active.
2. Do you think Jenna and Tasha are monozygotic or dizygotic twins? Why or why not?
3. What role might epigenetic influences play in determining Jenna and Tasha's development?

SUMMARY

2.1 Discuss the genetic foundations of development.
Genes are composed of stretches of deoxyribonucleic acid (DNA). 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, and others are recessive and will only be expressed if paired with another recessive gene. Other patterns include incomplete dominance and genomic imprinting. Most traits are polygenic, the result of interactions among many genes.

2.2 Identify examples of genetic disorders and chromosomal abnormalities.
Genetic disorders carried through dominant–recessive inheritance include PKU. Some recessive genetic disorders, such as hemophilia, are carried on the X chromosome. Males are more likely to be affected by X-linked genetic disorders. Fragile X syndrome is an example of a dominant–recessive disorder carried on the X chromosome. Other X-linked genetic disorders include Klinefelter syndrome, Jacob's syndrome, triple X syndrome, and Turner syndrome. Some disorders, such as Down syndrome, are the result of chromosomal abnormalities and others result from mutations. Individuals and couples turn to assisted reproductive technology (ART) for a variety of reasons, including reducing the risk of genetic or chromosomal abnormalities. ART includes artificial insemination, in vitro fertilization, and surrogacy.

2.3 Describe behavior genetics and the ways in which heredity and environment interact to influence development.
Behavior genetics investigates how genes and experience influence the diversity of human traits, abilities, and behaviors. Heritability research, including selective breeding studies,

family studies, and adoption studies, shows that genetics contributes to many traits. Passive, evocative, and active gene–environment correlations illustrate how traits are supported by both our genes and our environment. There is a wide range of potential expressions of a genetic trait, depending on environmental opportunities and constraints. Canalized traits 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.

2.4 Summarize three periods of prenatal development and commonly used prenatal diagnostic tests.

Conception occurs in the fallopian tube. During the germinal period, the zygote begins cell division and travels down the fallopian tube toward the uterus. During the embryonic period from weeks 2 to 8, the most rapid developments of the prenatal period take place. From 9 weeks until birth, the fetus grows rapidly, and the organs become more complex and begin to function. Ultrasound enables physicians to observe the fetus, measure fetal growth, judge gestational age, and detect physical abnormalities. Fetal MRI applies MRI technology to image the fetus's body and diagnose malformations. Amniocentesis and chronic villus sampling (CVS) involve extracting, growing, and analyzing a small sample of fetal cells from the amniotic fluid that surrounds the fetus. Noninvasive prenatal testing screens the mother's blood to detect chromosomal abnormalities, but it is not as accurate as amniocentesis or CVS.

2.5 Analyze the influence of teratogens on prenatal development.

Teratogens include diseases, drugs, and other agents that influence the prenatal environment to disrupt development. 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 abnormality, a variety of congenital abnormalities 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, smoking, and alcohol use can harm the developing fetus. The enforcement and consequences of prenatal substance use often vary with maternal demographic factors.

2.6 Examine the influence of parental characteristics and prenatal care on development.

Maternal influences on prenatal development include nutrition, health, and emotional well-being. Women who give birth over the age of 40 are at greater risk for preterm birth, miscarriage, and having a child with Down syndrome. Fathers' age and behavior may also influence the risk of chromosomal abnormalities and developmental disorders. Prenatal care is a set of services provided to improve pregnancy outcomes. There are significant racial, ethnic, and socioeconomic disparities in prenatal care that are thought to be largely influenced by socioeconomic factors. Although prenatal care predicts better birth outcomes, cultural factors, such as the Latina paradox, may protect some women and infants from the negative consequences of inadequate prenatal care.

2.7 Explain the process of childbirth.

Childbirth progresses through three stages: Uterine contractions cause the cervix to dilate; then the fetus passes through the birth canal, followed by the placenta. Medication is used in most births, often in combination with breathing and relaxation techniques

characteristic of natural births. Concerns about the health of the fetus or mother may prompt a cesarean delivery. Societies vary in their customs and perceptions of childbirth, including where it occurs and how newborns are integrated into the community.
The prevalence of low birthweight varies with ethnicity, socioeconomic status (SES), neighborhood, and access to resources such as prenatal care. Low-birthweight infants have difficulty adapting to their environment and are at risk for poor developmental outcomes. A warm stimulating home environment promotes positive outcomes.

KEY TERMS

age of viability (p. 80)
amniocentesis (p. 81)
amnion (p. 79)
anencephaly (p. 90)
Apgar scale (p. 99)
artificial insemination (p. 66)
assisted reproductive technology (ART) (p. 66)
behavior genetics (p. 68)
blastocyst (p. 77)
breech position (p. 97)
canalization (p. 70)
cesarean delivery (p. 97)
chorionic villus sampling (CVS) (p. 81)
chromosomes (p. 54)
deoxyribonucleic acid (DNA) (p. 54)
dizygotic (DZ) twin (p. 57)
dominant–recessive inheritance (p. 57)
Down syndrome (p. 63)
embryo (p. 78)
epigenetics (p. 74)
fetal alcohol spectrum disorders (p. 85)
fetal alcohol syndrome (FAS) (p. 85)
fetal MRI (p. 81)
fetal period (p. 79)
fetus (p. 79)
fragile X syndrome (p. 61)
gamete (p. 55)
gene (p. 54)
gene–environment correlation (p. 71)
gene–environment interactions (p. 69)
genomic imprinting (p. 59)
genotype (p. 57)
germinal period (p. 77)
hemophilia (p. 63)
heritability (p. 68)
implantation (p. 77)
in vitro fertilization (p. 66)
incomplete dominance (p. 57)
kangaroo care (p. 101)
Klinefelter syndrome (p. 64)
lanugo (p. 79)
low birthweight (p. 100)
meiosis (p. 55)
midwife (p. 98)
mitosis (p. 55)
monozygotic (MZ) twin (p. 57)
mutations (p. 65)
natural childbirth (p. 97)
neural tube (p. 79)
Niche picking (p. 72)
noninvasive prenatal testing (NIPT) (p. 81)
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prenatal care (p. 93)
range of reaction (p. 69)
sickle cell anemia (p. 57)
sleeper effects (p. 84)
small for date (p. 100)
spina bifida (p. 90)
surrogacy (p. 67)
teratogen (p. 83)
Turner syndrome (p. 64)
ultrasound (p. 80)
vernix caseosa (p. 80)
zygote (p. 55)