2 Biological and Environmental Foundations of Development

The average person cannot tell twin siblings apart because twins are virtually identical. Or are they? Consider these twin girls: Maria has strikingly dark hair and deep brown eyes, similar to her father. Maria's twin sister, Anna, seems to take after their mother, with blond hair and blue eyes. Maria and Anna not only differ in appearance, but they like different foods, have different interests, and have somewhat different personalities. Although Maria is more outgoing and sociable than Anna, they both enjoy spending quiet solitary time in their room reading, drawing, and daydreaming. As twins, Maria and Anna shared a womb and many early experiences. Shouldn't they be more similar? We tend to expect twins to share similarities, as they are often depicted in the media as identical in appearance, personality, and interests, yet they tend to differ in many unpredictable ways despite sharing parents and a home environment. Twins illustrate the complexity of how characteristics and tendencies are inherited. In this chapter, we discuss

Learning Objectives

2.1 Discuss the genetic foundations of development.

Video Activity 2.1: Twins

2.2 Identify examples of genetic disorders and chromosomal abnormalities.

2.3 Examine the choices available to prospective parents in having healthy children.

Video Activity 2.1: Genetics and Pregnancy

2.4 Summarize the interaction of heredity and environment, including behavioral genetics and the epigenetic framework.

Chapter Contents

the process of genetic inheritance and principles that can help us to understand how members of a family—even twins—can share a great many similarities and also many differences.

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. The following sections examine the role of heredity in our development.

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** (Plomin, DeFries, Knopik, & Neiderhiser, 2013). Each chromosome holds the basic units of heredity, known as **genes**, composed of stretches of **deoxyribonucleic acid (DNA)**, a complex molecule shaped like a twisted ladder or

staircase. Genes carry the plan for creating all of the traits that organisms carry. It is estimated that 20,000 to 25,000 genes reside within the chromosomes, comprising the human genome and influencing all genetic characteristics (Finegold, 2017).

Much of our genetic material is not unique to humans. Every species has a different genome, yet we share genes with all organisms, from bacteria to primates. We share 99% of our DNA with our closest genetic relative, the chimpanzee. There is even less genetic variation among humans. People around the world share 99.7% of their genes (Lewis, 2017). Although all humans share the same basic genome, every person has a slightly different code, making him or her genetically distinct from other humans.

Cell Reproduction

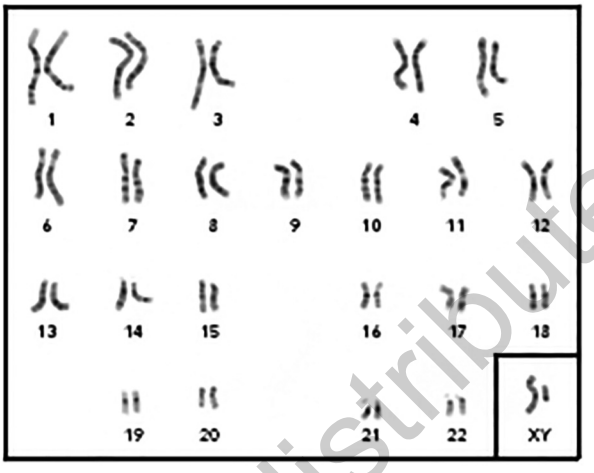
Most cells in the human body reproduce through a process known as **mitosis** in which DNA replicates itself, duplicating chromosomes, which ultimately form new cells with identical genetic material (Sadler, 2018). The process of mitosis accounts for the replication of all body cells. However, sex cells reproduce in a different way, 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 (Sadler, 2018). 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 it is estimated that individuals can produce millions of genetically different gametes (National Library of Medicine, 2019).

Sex Determination

Whether a zygote will develop into a male or female is controlled by the sex chromosomes. As shown in Figure 2.1, 22 of the 23 pairs of chromosomes are matched; they contain similar genes in almost identical positions and sequence, reflecting the distinct genetic blueprint of the biological mother and father. The 23rd pair of chromosomes are sex chromosomes that specify the biological sex of the individual. In females, sex chromosomes consist of two large X-shaped chromosomes (XX). Males' sex chromosomes consist of one large X-shaped chromosome and one much smaller Y-shaped chromosome (XY).

FIGURE 2.1

Chromosomes



Because females have two X sex chromosomes, all ova contain one X sex chromosome. A male's sex chromosome pair includes both X and Y chromosomes; therefore, one-half of the sperm males produce contains an X chromosome and one-half contains a Y. 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, as shown in Figure 2.2. (The introduction of sex selection methods has become more widely available, and some parents may seek to choose the sex of their child. For more on this topic, see the Applying Developmental Science feature.)

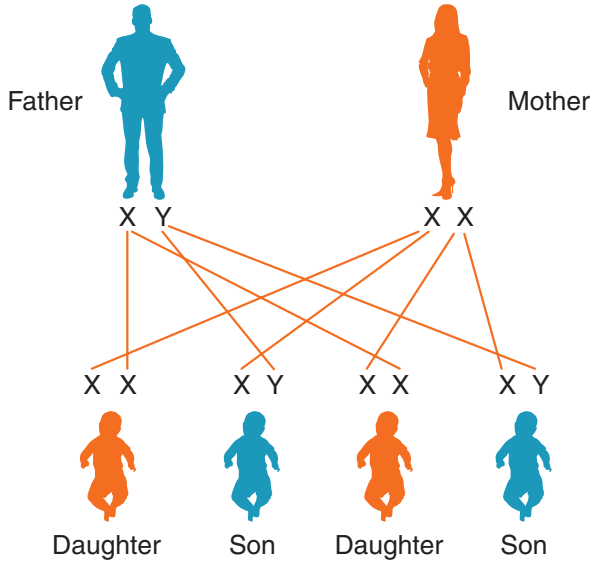
Genes Shared by Twins

All biological siblings share the same 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 (Martin, Hamilton, Osterman, Driscoll, & Drake, 2018).

The majority of naturally conceived twins are **dizygotic (DZ) twins**, or fraternal twins, conceived when a woman releases more than one ovum and each is fertilized by a different sperm. DZ twins share about one-half of their genes, and like other siblings, most fraternal twins differ in appearance,

FIGURE 2.2

Sex Determination

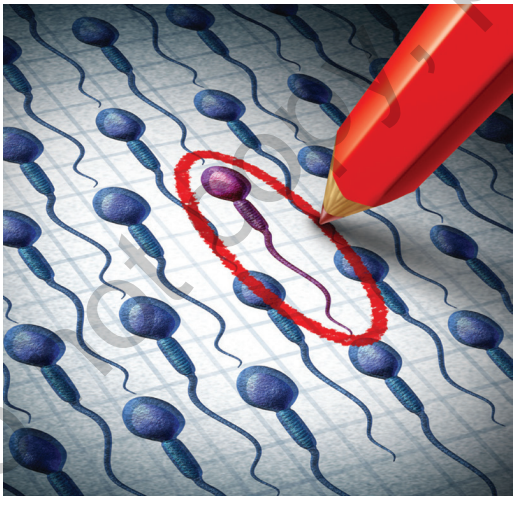


such as hair color, eye color, and height. In about half of fraternal twin pairs, one twin is a boy and the other a girl. DZ twins tend to run in families, suggesting a genetic component that controls the tendency for a woman to release more than one ovum each month. However, rates of DZ twins also increase with in vitro fertilization, maternal age, and each subsequent birth (Pison, Monden, & Smits, 2015; Umstad, Calais-Ferreira, Scurrah, Hall, & Craig, 2019).

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. It is estimated that MZ twins occur in 1 in every 250 births (Parazzini et al., 2016; Umstad et al., 2019). The causes of MZ twinning are not well understood. Rates of MZ twins are not related to maternal age or the number of births, but in vitro fertilization appears to increase the risk of MZ twins (Knopman et al., 2014; Umstad et al., 2019).

APPLYING DEVELOPMENTAL SCIENCE

Prenatal Sex Selection



Sperm cells can be sorted by whether they carry the X or Y chromosome. Through in vitro fertilization, a zygote with the desired sex is created.
Brain light/Alamy Stock Photo

Parents have long shown a preference for giving birth to a girl or boy, depending on circumstances such as cultural or religious traditions, the availability of males or females to perform certain kinds of work important to the family or society, or the sex of the couple's other children. Until recently, the sex of an unborn child was a matter of hope, prayer, and folk rituals. It is only in the past generation that science has made it possible for parents to reliably choose the sex of their unborn child. The introduction of sex selection has been a boon to couples carrying a genetically transmitted disease (i.e., a disease carried on the sex chromosomes), enabling them to have a healthy baby of the sex unaffected by the disease they carried.

Sex selection is generally conducted using two methods: preimplantation genetic diagnosis (PGD) or preconception sperm sorting (Bhatia, 2018). PGD creates zygotes within the laboratory by removing eggs from the woman and fertilizing them with sperm. This is known as in vitro (literally, "in glass") fertilization because fertilization takes place in a test tube, outside of the woman's body. After 3 days, a cell from the organism is used to examine the chromosomes and determine its sex. The desired male or female embryos are then implanted into the woman's uterus. PGD is generally conducted only when the risk of family genetic disorders is high and is about 99% effective.

(*Continued*)

(Continued)

Preconception sperm sorting entails spinning sperm in a centrifuge to separate those that carry an X or a Y chromosome. Because X sperm are denser than Y sperm, they are easily separated. Sperm with the desired chromosomes are then used to fertilize the ovum. Sperm sorting has been available and commonly used since the 1970s. The success rate is about 75% (Bhatia, 2018).

The availability of sex selection procedures enables parents to choose the sex of their child because of personal desires, such as to create family balance or to conform to cultural valuing of one sex over the other, rather than simply to avoid transmitting genetic disorders (Robertson & Hickman, 2013). Critics argue that sex selection can lead down a "slippery slope" of genetic engineering and selecting for other characteristics, such as appearance, intelligence, and more (Dondorp et al., 2013). Others express concerns about societal sex ratio imbalances if sex selection becomes widely practiced (Colls et al., 2009; Robertson & Hickman, 2013). Such sex ratio imbalances favoring males have occurred in India and China because of female infanticide, gender-driven abortion, and China's one-child family policy (see the Lives in Context: Cultural Context feature in Chapter 12 for more information; Bhatia, 2010, 2018).

Most Canadian, U.K., and European countries restrict the use of PGD and prohibit it for nonmedical reasons (Bayefsky, 2016). The United States does not have a formal policy regarding sex selection (Deeney, 2013). Sex selection remains hotly debated in medical journals, by hospital and university ethics boards, and by the public.

What Do You Think?

1. Should parents be able to choose the sex of their baby? Under what conditions is sex selection acceptable?
2. If you were able to selectively reproduce other characteristics, apart from sex, what might you choose? Why or why not? ●

Patterns of Genetic Inheritance

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

Dominant–Recessive Inheritance

Lynn has red hair but her brother, Jim, does not—and neither do their parents. How did Lynn end up with red hair? These outcomes can be explained by patterns of genetic inheritance, how the sets of genes from each parent interact. As we have discussed, each person has 23 pairs of chromosomes, one pair inherited from the mother and one from the father. The genes within each chromosome can be expressed in different forms, or **alleles**, that influence a variety of physical characteristics. When alleles of the pair of chromosomes are alike with regard to a specific characteristic, such as hair color, the person is said to be *homozygous* for the characteristic and will display the inherited trait. If they are different, the person is *heterozygous*, and the trait expressed will depend on the relations among the genes (Lewis, 2017). Some genes are passed through **dominant–recessive inheritance** in which some genes are *dominant* and are always expressed regardless of the gene they are paired with. Other genes are *recessive* and will be expressed only if paired with another recessive gene. Lynn and Jim's parents are heterozygous for red hair; both have dark hair, but they each carry a recessive gene for red hair.

When an individual is heterozygous for a particular trait, the dominant gene is expressed, and the person becomes a carrier of the recessive gene. For example, consider Figure 2.3. Both parents have nonred hair. People with nonred hair may have homozygous or heterozygous genes for hair color because the gene for nonred hair (symbolized by N in Figure 2.3) is dominant over the gene for red hair (r). In other words, both a child who inherits a homozygous pair of dominant genes (NN) and one who inherits a heterozygous pair consisting of both a dominant and recessive gene (Nr) will have nonred hair, even though the two genotypes are different. Both parents are heterozygous for red hair (Nr). They each carry the gene for red hair and can pass it on to their offspring. Red hair can result only from having two recessive genes (rr); both parents must carry the recessive gene for red hair. Therefore, a child with red hair can be born to parents who have nonred hair if they both carry heterozygous genes for hair color. As shown in Table 2.1, several characteristics are passed through 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	Colorblindness

Source: McKusick (1998) and McKusick-Nathans Institute of Genetic Medicine (2019).

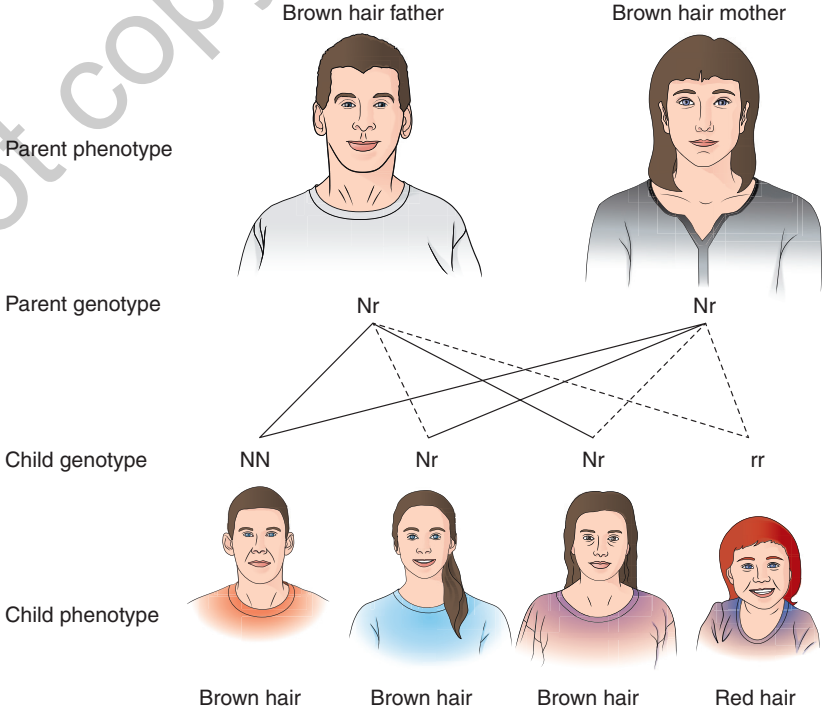
Incomplete Dominance

In most cases, dominant–recessive inheritance is an oversimplified explanation for patterns of genetic inheritance. **Incomplete dominance** is a genetic inheritance pattern in which both genes influence the characteristic (Finegold, 2017). For example, consider blood type. The alleles for blood types A and B do not dominate each other. A heterozygous person with the alleles for blood type A and B will express both A and B alleles and have blood type AB.

A different type of inheritance pattern is seen when a person inherits heterozygous alleles in which one allele is stronger than the other yet does not completely dominate. In this situation, the stronger allele does not mask all of the effects of the weaker allele. Therefore, some, but not all, characteristics of the recessive allele appear. For example, the trait for developing normal blood cells does not completely mask the allele for developing sickle-shaped blood cells. About 5% of African American newborns (and relatively few Caucasians or Asian Americans) carry the recessive **sickle cell trait** (Ojodu, Hulihan, Pope, & Grant, 2014). Sickle cell alleles cause red blood cells to become crescent, or sickle, shaped. Cells that are sickle shaped cannot distribute oxygen effectively throughout the circulatory system (Ware, de Montalembert, Tshilolo, & Abboud, 2017). The average life expectancy for individuals with sickle cell anemia is 55 years in North America (Pecker & Little, 2018). Alleles for normal blood cells do not mask

FIGURE 2.3

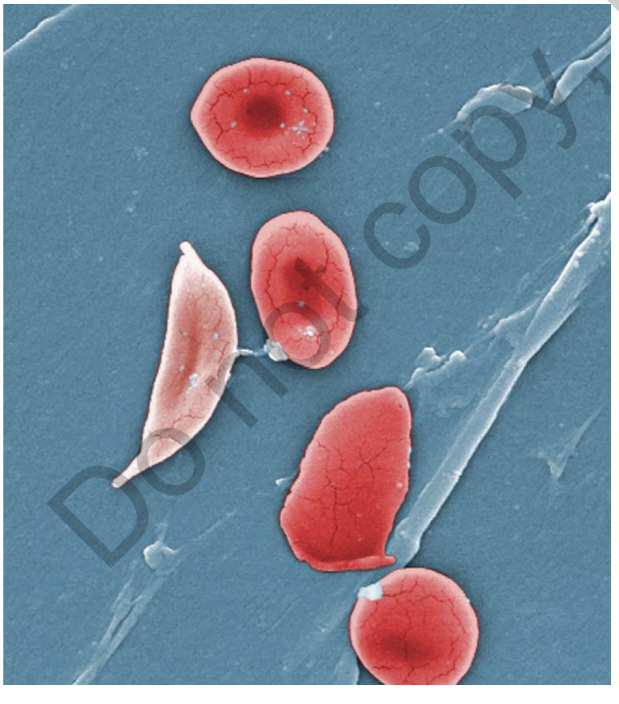
Dominant–Recessive Inheritance



all of the characteristics of recessive sickle cell alleles, illustrating incomplete dominance. Sickle cell carriers do not develop full-blown sickle cell anemia (Chakravorty & Williams, 2015). Carriers of the trait for sickle cell anemia may function normally but may show some symptoms such as reduced oxygen distribution throughout the body and exhaustion after exercise. Only individuals who are homozygous for the recessive sickle cell trait develop sickle cell anemia.

Polygenic Inheritance

Whereas dominant–recessive and codominant–recessive patterns account for some genotypes, most traits are a function of the interaction of many genes, known as **polygenic inheritance**. Hereditary influences act in complex ways, and researchers cannot trace most characteristics to only one or two genes. Instead, polygenic traits are the result of interactions among many genes. Examples of polygenic traits include height, intelligence, personality, and susceptibility to certain forms of cancer (Bouchard, 2014; Kremen, Panizzon, & Cannon, 2016; Penke & Jokela, 2016). 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.



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 (Amberger & Hamosh, 2017; McKusick, 2007). However, a few traits are determined by a process known as **genomic imprinting**. Genomic imprinting refers to the instance in which the expression of a gene is determined by whether it is inherited from the mother or the father (Kelly & Spencer, 2017; National Library of Medicine, 2019). For example, consider two conditions that illustrate genomic imprinting: Prader-Willi syndrome and Angelman syndrome. Both syndromes are caused by an abnormality in the 15th chromosome (Kalsner & Chamberlain, 2015). As shown in Figure 2.4, if the abnormality occurs on chromosome 15 acquired by the father, the individual—whether a daughter or son—will develop Prader-Willi syndrome, a set of specific physical and behavioral characteristics including obesity, insatiable hunger, short stature, motor slowness, and mild to moderate intellectual impairment (Butler, Manzardo, Heinemann, Loker, & Loker, 2016). If the abnormal chromosome 15 arises from the mother, the individual—again, whether a daughter or a son—will develop Angelman syndrome, characterized by hyperactivity, thin body frame, seizures, disturbances in gait, and severe learning disabilities, including severe problems with speech (Buiting, Williams, & Horsthemke, 2016). Prader-Willi and Angelman syndromes are rare, occurring on average in 1 in 12,000 to 20,000 persons (Kalsner & Chamberlain, 2015; Spruyt, Braam, & Curfs, 2018). Patterns of genetic inheritance can be complex, yet they follow predictable principles. For a summary of patterns of genetic inheritance, refer to Table 2.2.

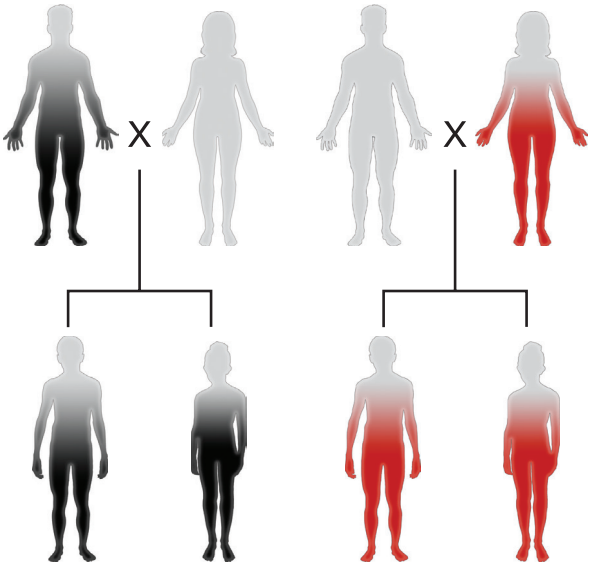
TABLE 2.2

Summary of Patterns of Genetic Inheritance

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

FIGURE 2.4

Genomic Imprinting



Source: C. Cristofre Martin (1998).

THINKING IN CONTEXT 2.1

1. Consider the evolutionary developmental perspective discussed in Chapter 1. From an evolutionary developmental perspective, why are some characteristics dominant and others recessive? Is it adaptive for some traits to dominate over others? Why or why not?
2. Consider your own physical characteristics, such as hair and eye color. Are they indicative of recessive traits or dominant ones? Which of your traits are likely polygenic?
3. From an evolutionary developmental perspective, why do twins occur? Do you think twinning serves an adaptive purpose? Explain.

CHROMOSOMAL AND GENETIC PROBLEMS

Many disorders are passed through genetic inheritance or are 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 over a period of many years.

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 such as sickle cell anemia, as well as others that are rare. Some are highly visible and others go unnoticed during an individual's life.

Dominant–Recessive Disorders

Recall that in dominant–recessive inheritance, dominant genes are always expressed regardless of the gene they are paired with and recessive genes are expressed only if paired with another recessive gene. Table 2.3 illustrates diseases that are inherited through dominant–recessive patterns. Few severe disorders are inherited through dominant inheritance because individuals who inherit the allele often do not survive long enough to reproduce and pass it to the next generation. One exception is Huntington disease, a fatal disease in which the central nervous system deteriorates (National Library of Medicine, 2019). Individuals with the Huntington allele develop normally in childhood, adolescence, and young adulthood. Symptoms of Huntington disease do not appear until age 35 or later. By then, many individuals have already had children, and one-half of them, on average, will inherit the dominant Huntington 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 (Kahn et al., 2016; Romani et al., 2017). Without treatment, the phenylalanine builds up quickly to toxic levels that damage the central nervous system, contributing to intellectual developmental disability, once known as mental retardation, by 1 year of age. The United States and Canada require all newborns to be screened for PKU (Blau, Shen, & Carducci, 2014).

PKU illustrates how genes interact with the environment to produce developmental outcomes. Intellectual disability results from the interaction of the genetic predisposition and exposure to phenylalanine from the environment (Blau, 2016). Children with PKU can process only very small amounts of phenylalanine. If the disease is discovered, the infant is placed on a diet low in phenylalanine. Yet it is very difficult to remove nearly all phenylalanine from the diet. Individuals who maintain a strict diet usually attain average levels of intelligence, although they tend to score lower than those without PKU (Jahja et al., 2017).

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 to 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 to 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 to 4,000 descendants of Central and Eastern European Jews	Recessive	Degenerative brain disease	None; most die by 4 years of age

Source: McKusick-Nathans Institute of Genetic Medicine (2019).

Some cognitive and psychological problems may appear in childhood and persist into adulthood, particularly difficulty in attention and planning skills, emotional regulation, depression, and anxiety (Hawks, Strube, Johnson, Grange, & White, 2018; Jahja et al., 2017). The emotional and social challenges associated with PKU, such as the pressure of a strict diet and surveillance from parents, may worsen these symptoms, and dietary compliance tends to decline in adolescence as young people push boundaries and seek independence (Medford, Hare, & Wittkowski, 2017).

A newborn's blood is tested for phenylketonuria (PKU), a genetic disorder in which the body lacks the enzyme that breaks down phenylalanine. Without treatment, the phenylketonuria builds up to toxic levels and can damage the central nervous system.
Marmaduke St. John/Alamy Stock Photo

X-Linked Disorders

A special instance of the dominant–recessive pattern occurs with genes that are located on the X chromosome (Shah, DeRemigis, Hageman, Sriram, & Waggoner, 2017). Recall that males (XY) have both an X and a Y chromosome. Some recessive genetic disorders, like the gene for red-green colorblindness, are carried on the X chromosome. Males are more likely to be affected by X-linked genetic disorders because they have only one X chromosome and therefore any genetic marks on their X chromosome are displayed. Females (XX) have two X chromosomes; a recessive gene located on one X chromosome will be masked by a dominant gene on the other X chromosome. Females are 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.

Hemophilia, a condition in which the blood does not clot normally, is another example of a recessive disease inherited through genes on the X chromosome (Shah et al., 2017). Daughters 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. 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. Sons who inherit the gene will display the disorder because the Y chromosome does not have the corresponding genetic information to counter the gene. Daughters who inherit the gene, again, will be carriers (unless their second X chromosome also carries the gene). Table 2.4 illustrates diseases acquired through X-linked inheritance.

In contrast, **fragile X syndrome** is an example of a dominant disorder carried on the X chromosome (Hagerman et al., 2017). Because the gene is dominant, it need appear on only one X chromosome to be displayed. That means that fragile X syndrome occurs in both males and females, although females tend to experience more mild symptoms. Males with fragile X syndrome typically have large ears, large testes, and a long, narrow face. Fragile X syndrome is the most common known inherited form of intellectual disability (Doherty & Scerif, 2017), and children with fragile X syndrome tend to show moderate to severe intellectual disability (Raspa, Wheeler, & Riley, 2017). Cardiac defects 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). Fragile X syndrome is often codiagnosed with autism, with estimates of 30% to 54% of boys and 16% to 20% of girls with fragile X syndrome meeting the diagnostic criteria for autism (Kaufmann et al., 2017).

Chromosomal Abnormalities

Chromosomal abnormalities are the result of errors during cell reproduction, meiosis or mitosis, or damage caused afterward. Occurring in about 1 of every 1,500 births, the most widely known chromosome disorder is trisomy 21, more commonly called **Down syndrome** (de Graaf, Buckley, Dever, & Skotko, 2017; Morrison & McMahon, 2018). Down syndrome occurs when a third chromosome appears alongside the 21st pair of chromosomes. Down syndrome is associated with marked physical, health, and cognitive attributes, including a short, stocky build, and striking facial features mark the disorder, such as a round face, almond-shaped eyes, and a flattened nose, as shown in Figure 2.5 (Davis & Escobar, 2013; Kruszka et al., 2017). Children with Down syndrome tend to show delays in physical and

TABLE 2.4

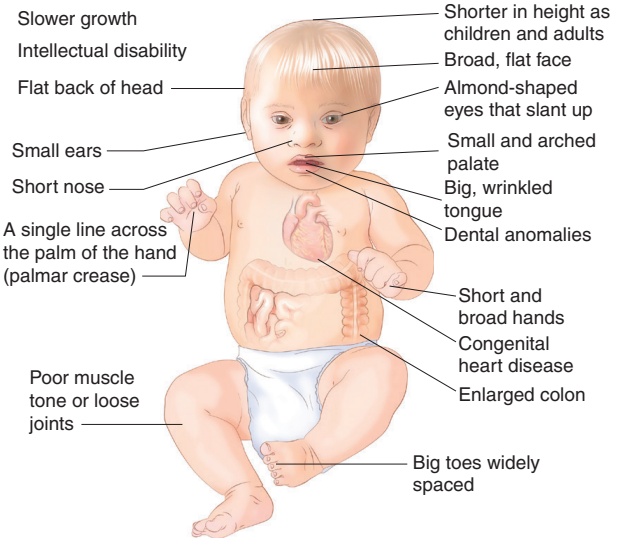
Diseases Acquired Through X-Linked Inheritance

SYNDROME/ DISEASE	OCCURRENCE	DESCRIPTION	TREATMENT
Colorblindness	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 to 7,000 males	Blood disorder in which the blood does not clot	Blood transfusions

Source: McKusick-Nathans Institute of Genetic Medicine (2019).

FIGURE 2.5

Down Syndrome



motor development relative to other children and health problems, such as congenital heart defects, vision impairments, poor hearing, and immune system deficiencies (Ram & Chinen, 2011; Zampieri et al., 2014). Down syndrome is the most common genetic cause of intellectual developmental disability (Vissers, Gilissen, & Veltman, 2016), but children's abilities vary. Generally, children with Down syndrome show greater strengths in nonverbal learning and memory relative to their verbal skills (Grieco, Pulsifer, Seligsohn, Skotko, & Schwartz, 2015). 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 emotion areas of functioning (Næss, Nygaard, Ostad, Dolva, & Lyster, 2017; Wentz, 2017).

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, compared with about 25 in the 1980s (Glasson, Dye, & Bittles, 2014; National Association for Down Syndrome, 2017). However, Down syndrome is associated with premature aging and an accelerated decline of cognitive functioning (Covelli, Raggi, Meucci, Paganelli, & Leonardi, 2016; Ghezzo et al., 2014). Individuals with Down syndrome are at risk to show signs of Alzheimer's disease very early relative to other adults (Hithersay, Hamburg, Knight, & Strydom, 2017; Wiseman et al., 2015). This is an example of how disorders and illnesses can be influenced by multiple genes and complex contextual interactions; in this case, Down syndrome and Alzheimer's disease share genetic markers (Lee, Chien, & Hwu, 2017).

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. They are summarized in Table 2.5.

Mutation

Not all inborn characteristics are inherited. Some result from **mutations**, sudden changes and abnormalities in the structure of genes that occur spontaneously or may be induced by exposure to environmental toxins such as radiation and agricultural chemicals in food (Lewis, 2017). A mutation may involve only one gene or many. It is estimated that as many as one-half of all conceptions include mutated chromosomes (Plomin et al., 2013). Most mutations are fatal—the developing organism 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. For example, 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). Children who inherited a single sickle cell allele were more resistant to malarial infection and more likely to survive and pass it along to their offspring (Croke et al., 2017; Gong, Parikh, Rosenthal, & Greenhouse, 2013). The sickle cell gene is not helpful in places of the world where malaria is not a risk. The frequency of the gene is decreasing in areas of the world where malaria is uncommon. For example, only 8% of African Americans are carriers, compared with as much as 30% of Black Africans in some African countries (Maakaron & Taher, 2012). Therefore, the developmental implications of genotypes—and mutations—are context specific, posing benefits in some contexts and risks in others. Recent advances in genetic engineering hold important implications for understanding and treating genetic disorders, as discussed in the Lives in Context feature.

TABLE 2.5

Sex Chromosome Abnormalities

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

Source: Ammerman et al. (2015); McKusick-Nathans Institute of Genetic Medicine (2019); Pappas and Migeon (2017); Wigby et al. (2016); and Wistuba, Brand, Zitzmann, and Damm (2017).

LIVES IN CONTEXT: BIOLOGICAL INFLUENCES

Genetic Engineering

We have seen that DNA influences many of our expressed traits. DNA, however, can be changed. Genetic engineering is a technology that permits scientists to change an organism's DNA. Genetic engineering is commonly used in agriculture and can be applied to modify plants to promote growth, strengthen crop resilience, and improve their nutritional value. In a process known as gene editing, genetic material can be added, removed, or changed at specific places on the genome (National Library of Medicine, 2019). One popular method of gene editing, CRISPR-Cas9 (commonly referred to as CRISPR), has generated a lot of excitement in the scientific community because it is faster, more cost-effective, more accurate, and more efficient than existing genome editing methods (National Library of Medicine, 2019).

Although gene editing is in its infancy, it is an experimental treatment for some genetic illnesses, including cancer and sickle cell anemia. Often referred to as gene therapy, the CRISPR method is used to manipulate the ill person's genome. At present, gene therapy is generally available only in research settings such as in experimental cancer treatment, for example

(*Continued*)

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(Stein, 2019). However, it holds promise. In recent clinical trials, some patients with sickle cell anemia have shown no signs of the disease after gene editing (Kolata, 2019).

Ethical concerns arise when gene editing is used to alter the human genome. Research on gene editing is limited to somatic cells, or body cells, because these changes affect only certain tissues and are not passed from generation to generation. Changes to sperm and egg cells, however, can be passed to future generations, posing ethical dilemmas such as the use of gene editing to enhance normal human traits. Because of these ethical issues as well as safety concerns, gene editing is not permitted on sperm and egg cells or embryos.

In late 2018, Chinese scientist He Jiankui garnered global media attention after using CRISPR to edit the DNA of two embryos. The embryos were implanted and carried to term by their mother. Jiankui's work was judged as unethical, and he was criticized for using untested, unregulated, and unsafe methods. In addition, the developmental consequences for the infants are unknown and any genetic abnormalities may be passed on to their kin. Jiankui argued that the infants' genomes were sequenced or mapped after birth, suggesting that only the intended genes were deleted. Nevertheless, the scientific community has condemned Jiankui's work and Jiankui is facing allegations of scientific misconduct and will also face criminal charges in his home country, China.

What Do You Think?

1. What are some of the pros and cons of genetic engineering?
2. What kinds of conditions should be treated by gene therapy? Are there any conditions that should be off limits to gene therapy? ●

THINKING IN CONTEXT 2.2

1. Give advice to prospective parents. Explain how genetic and chromosomal disorders are transmitted. What can parents do to reduce the risks?
2. Recall from Chapter 1 that most developmental scientists agree that nature and nurture 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.

REPRODUCTIVE CHOICES

The likelihood of genetic disorders often can be predicted before conception. Our growing understanding of genetic inheritance has led many couples to wonder about their own genetic inheritance and what genes they will pass on to their children.

Genetic Counseling

Many prospective parents seek **genetic counseling** to determine the risk that their children will inherit genetic defects and chromosomal abnormalities (Ioannides, 2017; Uhlmann, Schuette, & Yashar, 2009). Candidates for genetic counseling include those whose relatives have a genetic condition, couples who have had difficulties bearing children, women over the age of 35, and couples from the same ethnic group. Genetic testing can also determine whether a couple's difficulty conceiving or recurrent miscarriage is influenced by sperm chromosomal abnormalities in the male (Kohn, Kohn, Darilek, Ramasamy, & Lipshultz, 2016).

The genetic counselor interviews the couple to construct a family history of heritable disorders for both prospective parents. This service is particularly valuable when one or both prospective parents have relatives with inborn disorders. If a disorder is common in either parent's family or it appears that they are likely to carry a genetic disorder, genetic screening blood tests may be carried out on both parents to detect the presence of dominant and recessive genes and chromosomal abnormalities associated with various disorders. The tests determine whether each parent is a carrier for recessive disorders and estimate the likelihood that a child may be affected by a genetic disorder. The genetic counselor interprets the results and helps the parents understand genetic concepts by tailoring the explanation to match the parents' knowledge (Nance, 2017).

Once prospective parents learn about the risk of conceiving a child with a disorder, they can determine how to proceed—whether it is to conceive a child naturally or through the use of

in vitro fertilization—after screening gametes for the disorders of concern. Given advances in our knowledge of genetic disorders and the ability to screen for them, some argue that genetic counseling should be available to all prospective parents (Minkoff & Berkowitz, 2014). Others argue that abnormalities are rare and so few would be discovered that universal screening is of little utility (Larion, Warsof, Maher, Peleg, & Abuhamad, 2016). Whether to seek genetic counseling is a personal decision for prospective parents based on their history, view of their risks, and their values. Adults who carry significant risks of conceiving a child with a genetic disorder sometimes consider alternative methods of reproduction.

Assisted Reproductive Technology

Couples turn to assisted reproductive technology for a variety of reasons. As noted, some couples at risk for bearing children with genetic or chromosomal abnormalities seek alternative methods of conception. About 15% of couples in the United States experience infertility, the inability to conceive (Thoma et al., 2013). About 35% of the time, factors within the male are identified as contributors to infertility (Centers for Disease Control and Prevention, 2017c). In addition, single men and women, as well as gay and lesbian couples, often opt to conceive with the use of reproductive technology. However, there are racial, ethnic, and socioeconomic disparities in the use of assisted reproductive technologies. Women and couples who are White, college educated, and of high socioeconomic status (SES) are more likely to use infertility services than African American and Hispanic couples (Janitz, Peck, & Craig, 2016). Race and ethnicity are often linked with socioeconomic status and disparities in health care in the United States. Socioeconomic factors play a large role in access to infertility treatment and assisted reproductive technology (Dieke, Zhang, Kissin, Barfield, & Boulet, 2017).

One assisted reproduction technique is **artificial insemination**, the injection of sperm into a woman. The male partner's sperm may be used or, if the male experiences reproductive difficulties, a donor's sperm may be used. Artificial insemination through a donor also enables women without male partners, whether single or lesbian, to conceive. The most expensive assisted reproductive technology, in vitro fertilization, tends to average over $12,000 per trial, not including medication, and often requires multiple cycles, posing a financial burden too great for low SES women and couples (Teoh & Maheshwari, 2014).

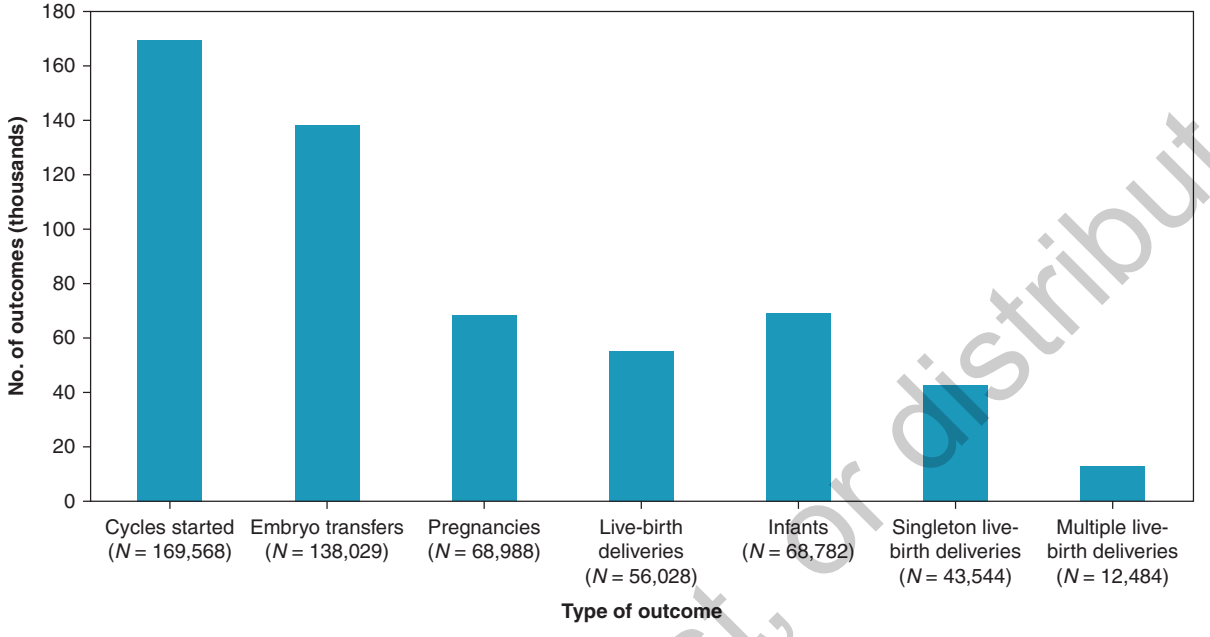
In vitro fertilization, introduced in the United States in 1981, permits conception to occur outside of the womb. A woman is prescribed hormones that stimulate the maturation of several ova, which are surgically removed. The ova are placed in a dish and sperm are added. One or more ova are fertilized, and the resulting cell begins to divide. After several cell divisions, the cluster of cells is 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. For example, the success rate is 47% in 35-year-old women, 27% in 41- to 42-year-old women, and 16% in 43- to 44-year-old women (Sunderam et al., 2017).

Assisted reproductive technology contributed to 1.6% of all infants born in the United States in 2014 (Sunderam et al., 2017). As shown in Figure 2.6, about 50% of assisted reproduction technology procedures that progress to the embryo-transfer stage result in pregnancy and about 40% result in a live birth. Infants conceived by in vitro fertilization are at higher risk of low birth weight (Fauser et al., 2014), although it has been suggested that it is because of maternal factors, such as advanced age, and not in vitro fertilization per se (Seggers et al., 2016). Infants conceived by in vitro fertilization show no differences in growth, health, development, and cognitive function relative to infants conceived naturally (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 birth defects (Fauser et al., 2014). However, about 40% of births from in vitro fertilization include more than one infant (38% twins and 2% triplets and higher). Multiple gestations increase risk for low birth weight, prematurity, and other poor outcomes (Sullivan-Pyke, Senapati, Mainigi, & Barnhart, 2017).

Surrogacy is an alternative form of reproduction 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 the child. 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. In 2015, 2,807 babies were born through surrogacy in the United States, up from 738 in 2004, according to the American Society for Reproductive Medicine (Beitsch, 2017). Longitudinal research suggests no psychological differences through age 14 between children born

FIGURE 2.6

Number of Outcomes of Assisted Reproductive Technology Procedures, by Type of Outcome—United States and Puerto Rico, 2014



Source: Sunderam et al. (2017).

through surrogacy compared with other methods, including children born to gay father and lesbian mother families (Carone, Lingiardi, Chirumbolo, & Baiocco, 2018; Golombok, 2013; Golombok, Ilioi, Blake, Roman, & Jadva, 2017). In addition, mothers of children who were the product of surrogates do not differ from those whose children were conceived using other methods, and surrogate mothers show no negative effects (Jadva, Imrie, & Golombok, 2015; Söderström-Anttila et al., 2015). Like other forms of reproductive technology, surrogacy is expensive, limiting its access to parents with high SES. Finally, some argue that surrogacy may pose ethical issues. For example, women are often paid at least $30,000 to surrogate a fetus (Beitsch, 2017), creating financial incentives that may be difficult for women with low SES to resist.

Adoption

Another reproductive option for prospective parents is **adoption**. Adults who choose to adopt have similar motives for parenthood as those who raise biological children, such as valuing family ties, continuing a family line, feeling that parenting is a life task, and desiring to have a nurturing relationship with a child (Jennings, Mellish, Tasker, Lamb, & Golombok, 2014; Malm & Welti, 2010). Heterosexual and same-sex adults report similar reasons for choosing adoption (Goldberg, Downing, & Moyer, 2012).

Adoptive children tend to be raised by parents with higher levels of education and income than other parents. This is partly due to self-selection and partly because of the screening that adoptive parents must undergo before they are allowed to adopt. It is estimated that transracial adoptions, in which a child (typically of color) is adopted by parents of a different race (most often White), account for about one-quarter of adoptions (Marr, 2017). Although there is little research, transracial adoptive children, and especially adolescents, may face challenges in ethnic and racial socialization and identity development (Wiley, 2017). Research reviews are mixed, with some suggesting no clear relation among racial or ethnic identity, parental socialization efforts, and adjustment (Boivin & Hassan, 2015) and more recent analyses suggesting that racial and ethnic socialization is associated with healthy adoptee outcomes (Montgomery & Jordan, 2018). Parents can foster their adoptive children's ethnic and racial socialization by

exposing children to their racial and ethnic heritage and providing opportunities for children to learn about and interact with people who identify with their birth race and ethnicity (Hrapczynski & Leslie, 2018).

Overall, adoptive children tend to spend more time with their parents and have more educational resources than other children (Zill, 2015). Yet some adopted children show less engagement in class and tend to have more academic difficulties than other children. Longitudinal research suggests that adoption is associated with lower academic achievement across childhood, adolescence, and emerging adulthood compared with nonadopted comparison groups (Brown, Waters, & Shelton, 2017). Adopted children tend to experience greater stress prenatally, early in life, prior to adoption, and during the adoption process that likely influences their long-term adjustment after adoption (Grotevant & McDermott, 2014). Adopted children therefore may show more psychological problems and adjustment difficulties than their nonadoptive peers, in some cases persisting into adulthood (A. Brown et al., 2017; Palacios & Brodzinsky, 2010).

Children's experiences prior to adoption and their developmental status at the time of adoption influence their outcomes (Balenzano, Coppola, Cassibba, & Moro, 2018). Children who experience neglect or fear and lack an early bond to a caregiver may experience difficulty regulating emotion and conflict. Biological mothers who choose to adopt may have experienced physical or mental health problems that interfered with their ability to care and form a bond and might be passed on. In other cases, the child may have experienced neglect, deprivation, and trauma, which influence adjustment (Grotevant & McDermott, 2014). Many children adopted from international orphanages arrive with experiences that are harmful, as discussed in the accompanying Lives in Context feature.

For many children, emotional differences are transitional. Research has suggested that most children show resilience in the years after adoption, but some issues continue (Palacios & Brodzinsky, 2010). Those who develop a close bond with adoptive parents tend to show better emotional understanding and regulation, social competence, and also self-esteem (Juffer & van IJzendoorn, 2007). This is true also of children who have experienced emotional neglect, and those effects hold regardless of age at adoption (Barone, Lionetti, & Green, 2017).

Prenatal Diagnosis

Prenatal testing is recommended when genetic counseling has determined a risk for genetic abnormalities, when the woman is older than age

LIVES IN CONTEXT: CULTURAL CONTEXT

Development of Internationally Adopted Children

Over the past 5 decades, international adoption has become commonplace. In many countries throughout the world, children are reared in orphanages with substandard conditions—without adequate food, clothing, or shelter and with poorly trained caregivers. Such orphanages have been found in a number of countries, including China, Ethiopia, Ukraine, Congo, and Haiti, accounting for over two-thirds of internationally adopted children (U.S. Department of State, 2014). Underfunded and understaffed orphanages often provide poor, nonnurturing care for children, increasing the risks for malnutrition, infections, physical disabilities, and growth retardation (Leiden Conference on the Development and Care of Children Without Permanent Parents, 2012). With high infant-to-caregiver ratios, children available for adoption often spend a significant amount of time deprived of consistent human contact.

Few internationally adopted children enter the United States healthy and at age-appropriate developmental norms. Not surprisingly, the longer the children are institutionalized, the more developmental challenges they face (Jacobs, Miller, & Tirella, 2010). Physical growth stunting is directly associated with the length of institutionalization, but catch-up growth is commonly seen after adoption (Wilson & Weaver, 2009). As with growth, the time spent in an orphanage predicts the degree of developmental delay. Longer institutionalization is associated with delays in the development of language, fine motor skills, social skills, attention, and other cognitive skills (Mason & Narad, 2005; Wiik et al., 2011).

Speech and language delays are among the most consistent deficiencies experienced by internationally

(*Continued*)

(Continued)

adopted children, especially those adopted after the age of 1 (Eigsti, Weitzman, Schuh, de Marchena, & Casey, 2011). However, more children reach normative age expectations 1 to 2 years postadoption (Glennen, 2014; Rakhlin et al., 2015). Generally, the younger the child is at adoption, the more quickly he or she will adapt to the new language and close any gaps in language delays (Glennan & Masters, 2002; Mason & Narad, 2005). Some research suggests that internationally adopted children are prone to long-term deficits in executive function likely due to neurological factors (Merz, Harlé, Noble, & McCall, 2016). The presence of a high-quality parent–child relationship promotes development of language, speech, or academic outcomes, and most children reach age-expected language levels (Glennen, 2014; Harwood, Feng, & Yu, 2013).

As adolescents, all children struggle to come to a sense of identity, to figure out who they are. This struggle may be especially challenging for internationally adopted children who may wonder about their native culture and homeland (Rosnati et al., 2015). Frequently, adolescents may want to discuss and learn more yet inhibit the desire to talk about this with parents (Garber & Grotevant, 2015). Parents who assume a multicultural perspective and provide opportunities for their children to learn about their birth culture support adopted children's development and promote healthy outcomes (Pinderhughes, Zhang, & Agerbak, 2015). Internationally adopted children seek to understand their birth culture and integrate their birth and adopted cultures into their sense of self (Grotevant, Lo, Fiorenzo, & Dunbar, 2017). A positive sense of ethnic identity is associated with positive outcomes such as self-esteem in international adoptees (Mohanty, 2015). Although there are individual differences in the degree of resilience and in functioning across developmental domains, adopted children overall show great developmental gains and resilience in physical, cognitive, and emotional development (Misca, 2014; Palacios, Román, Moreno, León, & Peñarrubia, 2014; Wilson & Weaver, 2009).

What Do You Think?

In your view, what are the most important challenges internationally adopted children and their families face? Identify sources and forms of support that might help adopted children and their parents. ●

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

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. Many deformities can be observed, such as cardiac abnormalities, cleft palate, and microencephaly (small head size). 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 to evaluate fetal development (Papp & Fekete, 2003). Repeated ultrasound of the fetus does not appear to affect growth and development (Stephenson, 2005).

Fetal MRI applies MRI technology to image the fetus's body and diagnose malformations (Griffiths et al., 2017). Most women will not have a fetal MRI. It is often used as a follow-up to ultrasound imaging to provide more detailed views of any suspected abnormalities (Milani et al., 2015). Fetal MRI can detect abnormalities throughout the body, including the central nervous system (Saleem, 2014). MRI in the obstetrical patient is safe for mother and fetus in the second and third trimesters but is expensive and has limited availability in some areas (Patenaude et al., 2014).

Amniocentesis is a prenatal diagnostic procedure in which a small sample of the amniotic fluid that surrounds the fetus is extracted from the mother's uterus through a long, hollow needle that is 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 and defects. Amniocentesis is less common than ultrasound, as it poses greater risk to the fetus. It is recommended for women aged 35 and older, 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 weeks of pregnancy. Conducted any earlier, an amniocentesis may increase the risk of miscarriage (Akolekar et al., 2015). Test results generally are available about

TABLE 2.6

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 onto 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.
Fetal MRI	Fetal MRI uses a magnetic scanner to record detailed images of fetal organs and structures.	Fetal MRI 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.
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 to create enough cells for genetic analysis.	Amniocentesis permits a thorough analysis of the fetus's genotype. There is a nearly 100% diagnostic success rate.	It is 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 fetus's genotype. CVS is relatively painless, and there is a 100% diagnostic success rate. It can be conducted earlier than amniocentesis, between 10 and 12 weeks.	It may pose a higher rate of spontaneous abortion and limb defects when conducted prior to 10 weeks' gestation.
Noninvasive prenatal testing (NIPT)	Cell-free fetal DNA is examined by drawing blood from the mother.	There is no risk to the fetus. NIPT 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, Beta, Picciarelli, Ogilvie, and D'Antonio (2015); Chan, Kwok, Choy, Leung, and Wang (2013); Gregg et al. (2013); Odibo (2015); Shahbazian, Barati, Arian, and Saadati (2012); Shim et al. (2014); and Theodora et al. (2016).

Ultrasound technology enables health care professionals to observe the fetus, measure fetal growth, detect physical abnormalities, and more.
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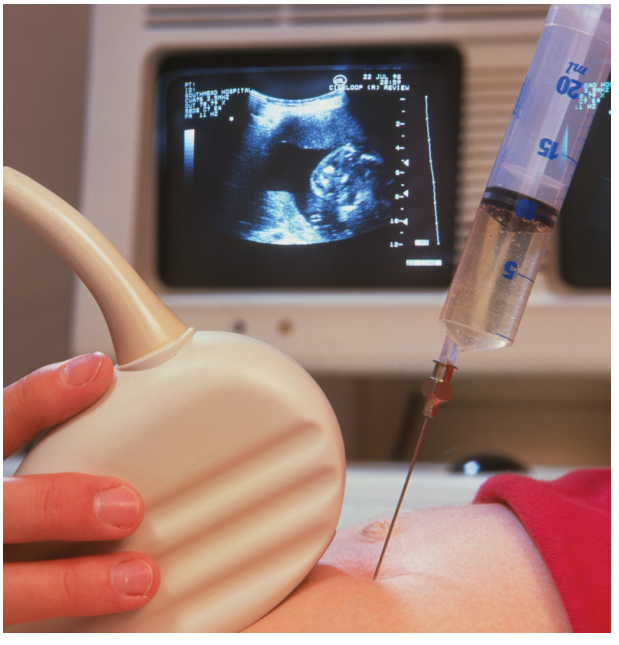
2 weeks after the procedure because it takes that long for the genetic material to grow and reproduce to the point where it can be analyzed.

Chorionic villus sampling (CVS) also samples genetic material and can be conducted earlier than amniocentesis, between 10 and 12 weeks of pregnancy (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 typically available about 1 week following the procedure. CVS is relatively painless and, like amniocentesis, has a 100% diagnostic success rate. Generally, CVS poses few risks to the fetus (Beta, Lesmes-



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

Heredia, Bedetti, & Akolekar, 2018; Shim et al., 2014). However, CVS should not be conducted prior to 10 weeks' gestation, as some studies suggest an increased risk of limb defects 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 in the breakdown of fetal cells) circulates in maternal blood in small concentrations that can be detected and studied by sampling the mother's blood (Warsof, Larion, & Abuhamad, 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, Angelotta, & Fisher, 2017). However, NIPT cannot detect as many chromosomal abnormalities as amniocentesis or CVS and does so with less accuracy (Chan et al., 2013; National Coalition for Health Professional Education in Genetics, 2012). 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). Pregnant women and their partners, in consultation with their obstetrician, should carefully weigh the risks and benefits of any procedure designed to monitor prenatal development.

Prenatal Treatment of Genetic Disorders

What happens when a genetic or chromosomal abnormality is found? Advances in genetics and in medicine have led to therapies that can be administered prenatally to reduce the effects of many genetic abnormalities. Fetoscopy is a technique that uses a small camera, inserted through a small incision on the mother's abdomen or cervix and placed into the amniotic sac that encases the fetus, to examine and perform procedures on the fetus during pregnancy. Risks of fetoscopy include infection, rupture of the amniotic sac, premature labor, and fetal death. However, when serious abnormalities are suspected, fetoscopy permits a visual assessment of the fetus, which aids in diagnosis and treatment. Hormones and other drugs, as well as blood transfusions, can be given to the fetus by inserting a needle into the uterus (Fox & Saade, 2012; Lindenburg, van Kamp, & Oepkes, 2014). Surgeons rely on the images provided by fetoscopy to surgically repair defects of the heart, lung, urinary tract, and other areas (Deprest et al., 2010; P. Sala et al., 2014).

In addition, researchers believe that one day, we may be able to treat many heritable disorders thorough genetic engineering by synthesizing normal genes to replace defective ones. It may someday be possible to sample cells from an embryo, detect harmful genes and replace them with healthy ones, and then return the healthy cells to the embryo where they reproduce and correct the genetic defect (Coutelle & Waddington, 2012). This approach has been used to correct certain heritable disorders in animals and holds promise for treating humans.

THINKING IN CONTEXT 2.3

1. 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?
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 tests? What would you advise? Why?

HEREDITY AND ENVIRONMENT

Our brief introduction to the processes of heredity illustrates the complexity of genetic inheritance. In fact, most human traits are influenced by a combination of genes (polygenic) working in concert with environmental influences. Our genotype**,** or genetic makeup, inherited from our biological parents is a biological contributor to all of our traits, from hair and eye color to personality, health, and behavior. However, our **phenotype**, the traits we ultimately show, such as our specific eye or hair color, is not determined by genotype, our genetic blueprint, alone. Phenotypes result from the interaction of genotypes and our experiences.

Behavioral Genetics

Behavioral genetics is the field of study that examines how genes and experience combine to influence the diversity of human traits, abilities, and behaviors (Krüger, Korsten, & Hoffman, 2017; Plomin et al., 2013). Behavioral geneticists have discovered that even traits with a strong genetic component, such as height, are modified by environmental influences (Dubois et al., 2012; Plomin, DeFries, Knopik, & Neiderhiser, 2016). Moreover, most human traits, such as intelligence, are influenced by multiple genes, and there are often multiple variants of each gene and each might interact with the environment in a different way (Bouchard, 2014; Chabris, Lee, Cesarini, Benjamin, & Laibson, 2015; Knopik, Neiderhiser, DeFries, & Plomin, 2017).

Methods of Behavioral Genetics

Behavioral 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 not due to genetic differences is instead a result of the environment and experiences. Heritability research therefore examines the contributions of the genotype but also provides information on the role of experience in determining phenotypes (Plomin et al., 2016). Behavioral geneticists assess the hereditary contributions to behavior by conducting selective breeding and family studies.

Selective breeding studies entail deliberately modifying the genetic makeup of animals to examine the influence of heredity on attributes and behavior. For example, mice can be bred to be very physically active or sedentary by mating highly active mice only with other highly active mice and, similarly, by breeding mice with very low levels of activity with each other. Over subsequent generations, mice bred for high levels of activity become many times more active than those bred for low levels of activity (Knopik et al., 2017). Selective breeding in rats, mice, and other animals such as chickens has revealed genetic contributions to many traits and characteristics, such as aggressiveness, emotionality, sex drive, and even maze learning (Plomin et al., 2016).

For many reasons, especially ethical reasons, people cannot be selectively bred. However, we can observe people who naturally vary in shared genes and environment. Behavioral geneticists conduct *family studies* to compare people who live together and share varying degrees of relatedness. Two kinds of family studies are common: twin studies and adoption studies (Koenen, Amstadter, & Nugent, 2012). *Twin studies* compare identical and fraternal twins to estimate how much of a trait or behavior is attributable to genes. 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 from 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.

Adoption studies, on the other hand, compare the degree of similarity between adopted children and their biological parents whose genes they share (50%) and their adoptive parents with whom they share an environment but not genes. If the adopted children share similarities with their biological parents, even though they were not raised by them, it suggests that the similarities are genetic. The similarities are influenced by the environment if the children are more similar to their adoptive parents. Observations of adoptive siblings also shed light on the extent to which attributes and behaviors are influenced by the environment. For example, the degree to which two genetically unrelated adopted children reared together are similar speaks to the role of environment. Comparisons of identical twins reared in the same home with those reared in different environments can also illustrate environmental contributions to phenotypes. If identical twins reared together are more similar than those reared apart, an environmental influence can be inferred.

Genetic Influences on Personal Characteristics

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

Genes contribute to many other traits, such as sociability, 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 (Esposito et al., 2017; McRae et al., 2017; Ritz et al., 2017). Yet even traits that are thought to be heavily influenced by genetics can be modified by physical and social interventions. For example, growth, body weight, and body height are largely predicted by genetics, yet environmental circumstances and opportunities influence whether genetic potentials are realized (Dubois et al., 2012; Jelenkovic et al., 2016). Even identical twins who share 100% of their genes are not 100% alike. Those differences are due to the influence of environmental factors, which interact with genes in a variety of ways.

TABLE 2.7

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

	REARED TOGETHER	REARED APART
Monozygotic twins (100% shared genes)	.86	.72
Dizygotic 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)[a]	.34	—
Nonbiological parent/child (0% shared genes)[a]	.19	—

Source: Adapted from Bouchard and McGue (1981).

[a] Estimated correlation for individuals sharing neither genes nor environment = .0.

Gene–Environment Interactions

We have seen that genes and the environment work together in complex ways to determine our characteristics, behavior, development, and health (Chabris et al., 2015; Ritz et al., 2017; Rutter, 2012). **Gene–environment interactions** refer to the dynamic interplay between our genes and our environment. Several principles illustrate these interactions.

Range of Reaction

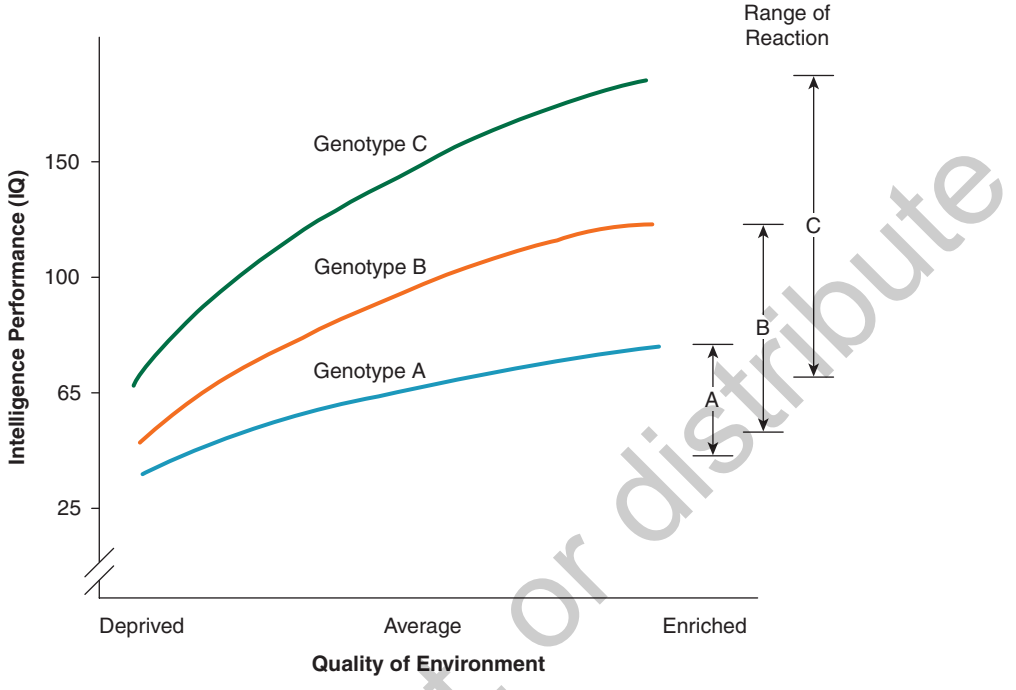
Everyone has a different genetic makeup and therefore responds to the environment in a unique way. In addition, any one genotype can be expressed in a variety of phenotypes. There is a **range of reaction** (see Figure 2.7), a wide range of potential expressions of a genetic trait, depending on environmental opportunities and constraints (Gottlieb, 2000, 2007). For example, consider height. Height is largely a function of genetics, yet an individual may show a range of sizes depending on environment and behavior. Suppose that a child is born to two very tall parents. She may have the genes to be tall, but unless she has adequate nutrition, she will not fulfill her genetic potential for height. In societies in which nutrition has improved dramatically over a generation, it is common for children to tower over their parents. The enhanced environmental opportunities (in this case, nutrition) enabled the children to fulfill their genetic potential for height. Therefore, a genotype sets boundaries on the range of possible phenotypes, but the phenotypes ultimately displayed vary in response to different environments (Manuck & McCaffery, 2014). In this way, genetics sets the range of development outcomes and the environment influences where, within the range, that person will fall.

Canalization

Some traits illustrate a wide reaction range. Others are examples of **canalization**, in which heredity narrows the range of development to only one or a few outcomes. Canalized traits are biologically programmed, and only powerful environmental forces can change their developmental path (Flatt, 2005; Posadas & Carthew, 2014; Waddington, 1971).

FIGURE 2.7

Range of Reaction



Source: Adapted from Gottlieb (2007).

For example, infants follow an age-related sequence of motor development, from crawling, to walking, to running. Around the world, most infants walk at about 12 months of age. Generally, only extreme experiences or changes in the environment can prevent this developmental sequence from occurring. For example, children reared in impoverished international orphanages and exposed to extreme environmental deprivation demonstrated delayed motor development, with infants walking 5 months to a year later than expected (Chaibal, Bennett, Rattanathanthong, & Siritaratiwat, 2016; Miller, Tseng, Tirella, Chan, & Feig, 2008).

Motor development is not entirely canalized, however, because some minor changes in the environment can subtly alter its pace and timing. For example, practice facilitates stepping movements in young infants, prevents the disappearance of stepping movements in the early months of life, and leads to an earlier onset of walking (Adolph & Franchak, 2017; Ulrich, Lloyd, Tiernan, Looper, & Angulo-Barroso, 2008). These observations demonstrate that even highly canalized traits, such as motor development, which largely unfolds via maturation, can be subtly influenced by contextual factors.

Gene–Environment Correlations

Heredity and environment are powerful influences on development. Not only do they interact, but environmental factors often support hereditary traits (Plomin et al., 2016; 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 (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.

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 also correspond to their child's genotype—an example of a *passive gene-environment correlation* (Wilkinson, Trzaskowski, Haworth, & Eley, 2013). It is a passive gene–environment correlation because it occurs regardless of the child's behavior. For example, a parent might provide genes that predispose a child to develop music ability and create a home environment that reflects the parent's interest

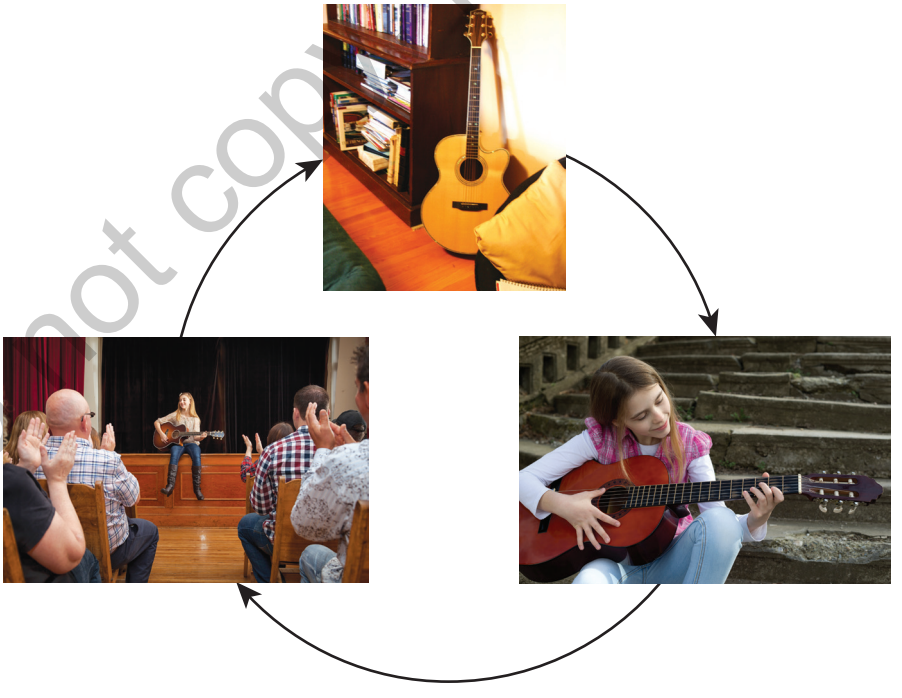
and ability in music, which then also happens to support the child's musical ability, as shown in the top photo in Figure 2.8. This type of gene–environment correlation tends to occur early in life because parents create rearing environments for their infants and young children.

Children naturally evoke responses from others and the environment, just as the environment and the actions of others evoke responses from the individual. In an *evocative gene–environment correlation*, a child's genetic traits (e.g., personality characteristics, including openness to experience) influence the social and physical environment, which in turn shape development in ways that support the genetic trait (Burt, 2009; Klahr, Thomas, Hopwood, Klump, & Burt, 2013). For example, active, happy infants tend to receive more adult attention than do passive or moody infants (Deater-Deckard & O'Connor, 2000), and even among infant twins reared in the same family, the more outgoing and happy twin receives more positive attention than does the more subdued twin (Deater-Deckard, 2001). Why? Babies who are cheerful and smile often influence their social world by evoking smiles from others, which in turn supports the tendency to be cheerful. In this way, genotypes influence the physical and social environment to respond in ways that support the genotype. Children who engage in disruptive play tend to later experience problems with peers (Boivin et al., 2013). To return to the music example, a child with a genetic trait for music talent will evoke pleasurable responses (e.g., parental approval) when she plays music; this environmental support, in turn, encourages further development of the child's musical trait. In addition, individuals vary in their sensitivity to environmental stimuli; some children may be more affected by environmental stimuli due to their genetic makeup (Belsky & Hartman, 2014; Pluess, 2015).

Children also take a hands-on role in shaping their development. Recall from Chapter 1 that a major theme in understanding human development is the finding that individuals are active in their development. Here we have an example of this theme. As children grow older, they have increasing freedom in choosing their own activities and environments. An *active gene–environment correlation* occurs when the child actively creates experiences and environments that correspond to and influence his genetic predisposition. For

FIGURE 2.8

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).
iStock/Essentials; iStock/Signature

example, the child with a genetic trait for interest and ability in music actively seeks experiences and environments that support that trait, such as friends with similar interests and after-school music classes. This tendency to actively seek out experiences and environments compatible with and supportive of our genetic tendencies is called **niche-picking** (Corrigall & Schellenberg, 2015; Scarr & McCartney, 1983).

The strength of passive, evocative, and active gene–environment correlations changes with development, as shown in Figure 2.9 (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 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, as well as select similar spouses and best friends (McGue & Christensen, 2013; Plomin & Deary, 2015; Plomin et al., 2016; Rushton & Bons, 2005).

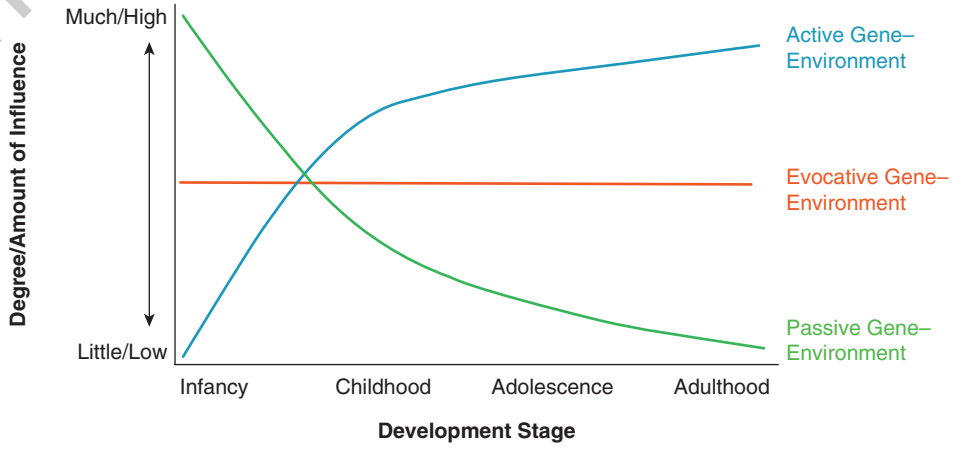
Epigenetic Influences on Development

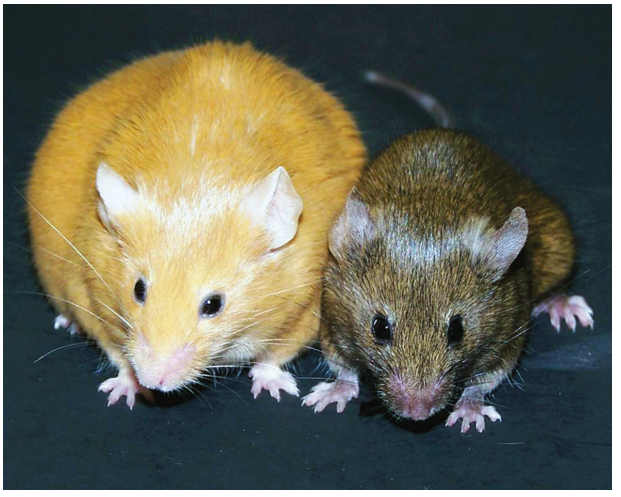
We have seen that development is influenced by the dynamic interaction of biological and contextual forces. Genes provide a blueprint for development, but phenotypic outcomes, individuals' characteristics, are not predetermined. Our genes are expressed as different phenotypes in different contexts or situations, known as epigenetics (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, for example. 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, Conradt, & Marsit, 2016). Environmental factors such as toxins, injuries, crowding, diet, and responsive parenting can influence the expression of genetic traits. In this way, even traits that are highly canalized can be influenced by the environment.

FIGURE 2.9

Development Stage and Gene–Environment Correlations





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

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). The mice in the photo both carry the agouti gene, yet they look very different; the brown mouse is slender and 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 it is turned on all the time in the yellow mouse and turned off in the brown mouse.

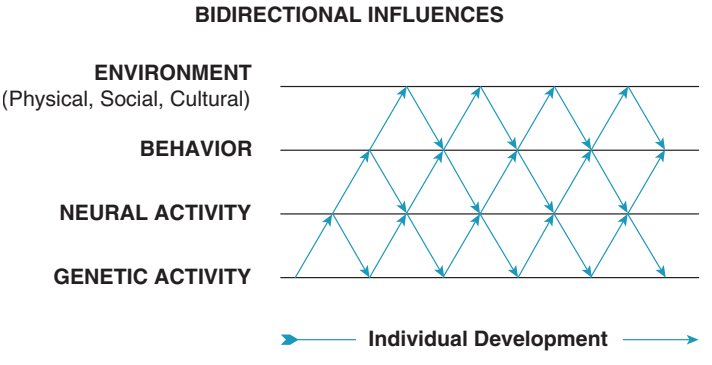
In 2003, Waterland and Jertle discovered that the pregnant agouti female's diet can determine her offspring's phenotype. In this study, female mice were fed foods containing chemicals that attach to a gene and turn it off. Yellow agouti mothers fed extra nutrients passed along the agouti gene to their offspring, but it was turned off. The mice looked radically different from their mother (brown) and were healthier (lean, not susceptible to disease) even though they carried the same genes.

Epigenetic processes also influence human development. For example, consider brain development. Providing an infant with a healthy diet and opportunities to explore the world will support the development of brain cells, governed by epigenetic mechanisms that switch genes on and off. Brain development influences motor development, further supporting the infant's exploration of the physical and social world and thereby promoting cognitive and social development. Active engagement with the world encourages connections among brain cells. Conversely, epigenetic changes that accompany exposure to toxins or extreme trauma might suppress the activity of some genes, potentially negatively influencing brain development and its cascading effects on motor, cognitive, and social development. In this way, an individual's neurological capacities are the result of epigenetic interactions among genes and contextual factors that determine his or her phenotype (Lerner & Overton, 2017). These complex interactions are illustrated in Figure 2.10 (Dodge & Rutter, 2011). 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).

Perhaps the most surprising finding emerging from animal studies of epigenetics, however, is that not only can the epigenome be influenced by the environment before birth but it can be passed by males and females from one generation to the next without changing the DNA itself (Soubry, Hoyo, Jirtle, & Murphy, 2014; Szyf, 2015). This means that what you eat and do today could affect the epigenome—the development, characteristics, and health—of your children, grandchildren, and great-grandchildren (Bale, 2015; Vanhees, Vonhögen, van Schooten, & Godschalk, 2014).

FIGURE 2.10

Epigenetic Framework



Source: Gottlieb (2007). With permission from John Wiley & Sons.

THINKING IN CONTEXT 2.4

1. Describe a skill or ability in which you excel. How might your ability be influenced by your genes and your context?
   a. Identify passive gene–environment correlation that may contribute to your ability. How has your environment influenced your ability?
   b. Provide an example of an evocative gene–environment correlation. How have you evoked responses from your context that influenced your ability?
   c. Explain how your ability might reflect an active gene–environment correlation.
   d. Which of these types of gene–environment correlations do you think best accounts for your ability? Why?
2. Considering the research on epigenetics, what can you do to protect your epigenome? What kinds of behavioral and contextual factors might influence your epigenome?

APPLY YOUR KNOWLEDGE

Zennia is sitting in her doctor's office. She tells Dr. Rasheed, "I want to have a baby. I have no partner, but I'm ready. I'm in my late 30s and financially stable. It's time. What are my options?" Dr. Rasheed replies, "There are a number of choices. It's a matter of figuring out what's right for you. In addition to a full examination to assess your health, we will seek assistance from a genetic counselor to determine the risk for genetic disorders. This information can help you decide among reproductive options."

1. Identify three ways that genetic disorders are passed. Why does Dr. Rasheed advise genetic testing?
2. What are some of the reproductive options available to Zennia? What are some of the advantages and disadvantages of each option?
3. Which option do you suggest? Why?
4. Suppose that Zennia became pregnant. What types of prenatal screening tests might Dr. Rasheed prescribe? Discuss some of the advantages and disadvantages of each.
5. What advice would you give Zennia? Why?

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KEY TERMS

Chromosome 35
Gene 35
Deoxyribonucleic acid (DNA) 35
Mitosis 36
Meiosis 36
Gamete 36
Zygote 36
Dizygotic (DZ) twins 36
Monozygotic (MZ) twins 37
Genotype 37
Allele 38
Dominant–recessive inheritance 38
Incomplete dominance 39
Sickle cell trait 39
Polygenic inheritance 40
Genomic imprinting 40
Phenylketonuria (PKU) 41
Hemophilia 43
Fragile X syndrome 43
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Mutation 44
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SUMMARY

2.1 Discuss the genetic foundations of development.

The human body is composed of trillions of units called cells, each with a nucleus containing 23 matching pairs of chromosomes, which contain genes, composed of stretches of deoxyribonucleic acid (DNA) that carry the plan for creating all of the traits that organisms carry. Some genes are passed through dominant–recessive inheritance, in which some genes are dominant and will always be expressed regardless of the gene they are paired with. Other genes are recessive and will only be expressed if paired with another recessive gene. When a person is heterozygous for a particular trait, the dominant gene is expressed and the person remains a carrier of the recessive gene. Some traits are carried on the X chromosome. Some traits are passed through incomplete dominance in which both genes influence the characteristic. Polygenic traits are the result of interactions among many genes. Some traits are determined by genomic imprinting, determined by whether it is inherited from the mother or the father.

2.2 Identify examples of genetic disorders and chromosomal abnormalities.

Genetic disorders carried through dominant–recessive inheritance include phenylketonuria (PKU), a recessive disorder, and Huntington disease, carried by a dominant allele. Some recessive genetic disorders, like the gene for hemophilia, are carried on the X chromosome. Males are more likely to be affected by X-linked genetic disorders, such as hemophilia. Fragile X syndrome is an example of a dominant–recessive disorder carried on the X chromosome. Because the gene is dominant, it must appear on only one X chromosome to be displayed. Other X-linked genetic disorders include Klinefelter syndrome, Jacob's syndrome, triple X syndrome, and Turner syndrome. Some disorders, such as trisomy 21, known as Down syndrome, are the result of chromosomal abnormalities. Others result from mutations, genetic abnormalities that may occur randomly or as a result of exposure to toxins.

2.3 Examine the choices available to prospective parents in having healthy children.

Genetic counseling is a medical specialty that helps prospective parents determine the likelihood that their children will inherit genetic defects and chromosomal abnormalities. Single women, gay and lesbian couples, and individuals at risk for bearing children with genetic or chromosomal abnormalities may seek alternative methods of conception such as artificial insemination, in vitro fertilization, and surrogacy. Others consider adopting a child. Prenatal diagnosis is recommended when genetic testing has determined a risk for genetic abnormalities. Some prenatal tests, such as ultrasound, are conducted routinely. Advances in genetics and in medicine have led to therapies that can be administered prenatally to reduce the effects of many genetic abnormalities.

2.4 Summarize the interaction of heredity and environment, including behavioral genetics and the epigenetic framework.

Behavioral genetics is the field of study that examines how genes and experience combine to influence the diversity of human traits, abilities, and behaviors. Heritability research examines the contributions of the genotype in determining phenotypes but also provides information on the role of experience through three types of studies: selective breeding studies, family studies, and adoption studies. Genetics contributes to many traits, such as intellectual ability, sociability, anxiety, agreeableness, activity level, obesity, and susceptibility to various illnesses. Passive, evocative, and active gene–environment correlations illustrate how traits often are supported by both our genes and environment. Reaction range refers to the idea that there is a wide range of potential expressions of a genetic trait, depending on environmental opportunities and constraints. Some traits illustrate canalization and require extreme changes in the environment to alter their course. The epigenetic framework is a model for understanding the dynamic ongoing interactions between heredity and environment whereby the epigenome's instructions to turn genes on and off throughout development are influenced by the environment.

REVIEW QUESTIONS

2.1 What genes are shared by twins?

What are four types of genetic inheritance?

2.2 Give an example of the following:

- a dominant–recessive disorder
- an X-linked disorder
- a chromosomal abnormality

What is imprinting?

What is a mutation?

2.3 What is genetic counseling?

What are the differences between artificial insemination, in vitro fertilization, surrogacy, and adoption?

What are the five common methods of prenatal diagnosis?

2.4 What is behavioral genetics?

What are three types of gene—environment correlations?

What is the range of reaction?

What is the epigenetic framework?

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