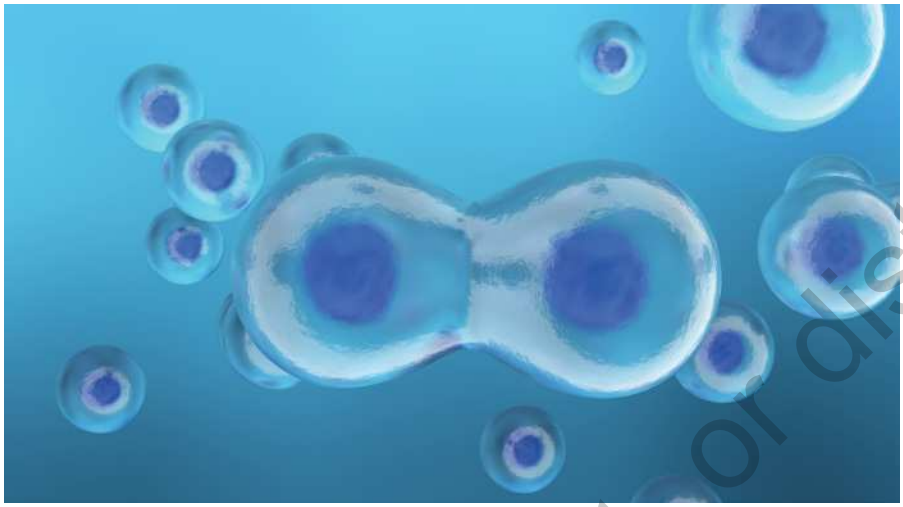
2 BIOLOGICAL AND ENVIRONMENTAL FOUNDATIONS OF DEVELOPMENT



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"Rico and Remmy couldn't be more different," marveled their mother. "People are surprised to find out they are twins." Rico is tall and athletic, with blond hair and striking blue eyes. He spends most afternoons playing ball with his friends and often invites them home to play in the yard. Remmy is much smaller, thin and wiry. He wears thick glasses over his brown eyes that are nearly as dark as his hair. Unlike his brother, Remmy prefers solitary games and spends most afternoons at home playing video games, building model cars, and reading comic books. How can Rico and Remmy share the same womb, have the same parents, and live in the same home yet differ markedly in appearance, personality, and preferences?

We tend to expect twins like Rico and Remmy to share similarities, as they are often depicted in media as identical in appearance, personality, and interests. Yet twins 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 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

LEARNING OBJECTIVE

2.1 Discuss the 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. This blueprint is inherited from our biological parents, as discussed in the following sections.

Genetics

The human body is composed of trillions of units called cells, each with a nucleus containing 23 matching pairs of rod-shaped structures called **chromosomes** (Finegold, 2019). Each chromosome holds the basic units of heredity, known as genes, composed of stretches of **deoxyribonucleic acid (DNA)**, a complex molecule shaped like a twisted ladder or staircase. **Genes** carry the plan for creating all of the traits that organisms carry. It is estimated that 20,000 to 25,000 genes reside within the chromosomes, comprising the human genome and influencing all genetic characteristics (Taneri et al., 2020).

Much of our genetic material is not unique to humans. Every species has a different genome, yet we share some genes with all organisms, from bacteria to primates. We share nearly 99% of our DNA with our closest genetic relative, the chimpanzee. There is even less genetic variation among humans. People around the world share 99.9% of their genes (Lewis, 2017; National Human Genome Research Institute, 2018). Although all humans share the same basic genome, every person has a slightly different code, making them genetically distinct from other humans.

Cell Reproduction

Most cells in the human body reproduce through a process known as **mitosis,** in which DNA replicates itself, duplicating chromosomes, resulting in new cells with identical genetic material (Sadler, 2018). The process of mitosis accounts for the replication of body cells. 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 (Finegold, 2019). The resulting cell consists of only 23 single, unpaired chromosomes. Known as **gametes**, these cells are specialized for sexual reproduction: sperm in males and ova in females. Ova and sperm join at fertilization to produce a fertilized egg, or **zygote**, with 46 chromosomes, forming 23 pairs with half from the biological mother and half from the biological father. Each gamete has a unique genetic profile, and it is estimated that individuals can produce millions of genetically different gametes (U.S. National Library of Medicine, 2020).

Sex Determination

The sex chromosomes determine whether a zygote will develop into a male or female. Twenty-two of the 23 pairs of chromosomes are matched pairs (Figure 2.1). They contain similar genes in almost identical positions and sequence, reflecting the distinct genetic blueprint of the biological mother and father. The 23rd pair of chromosomes are not identical because they are sex chromosomes that specify the genetic sex of the individual. In females, sex chromosomes consist of two large X-shaped chromosomes (XX). Males' sex chromosomes consist of one large X-shaped chromosome and one much smaller Y-shaped chromosome (XY).

Because females have two X sex chromosomes, all their ova contain one X sex chromosome. A male's sex chromosome pair includes both X and Y chromosomes; therefore, one half of the sperm males produce contain an X chromosome and one half contain a Y. The Y chromosome contains genetic instructions that will cause the fetus to develop male reproductive organs. Thus, whether the fetus develops into a boy or girl is determined by which sperm fertilizes the ovum. If the ovum is fertilized by a Y sperm, a male fetus will develop, and if the ovum is fertilized by an X sperm, a female fetus will form (Figure 2.2).

Genes Shared by Twins

All biological siblings share the same 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 et al., 2018).

The majority of naturally conceived twins (over 70%) are dizygotic (DZ) twins, or fraternal twins, conceived when a woman releases more than one ovum and each is fertilized by a different sperm (Gill et al., 2019). DZ twins share about one half of their genes and, like other siblings, most fraternal twins

FIGURE 2.1 ■ Chromosomes

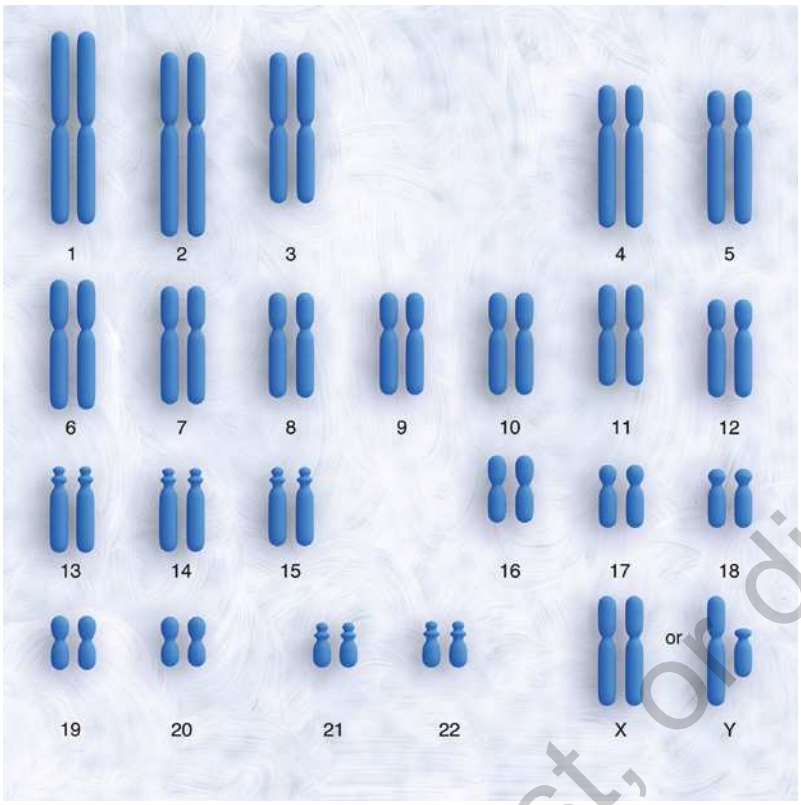

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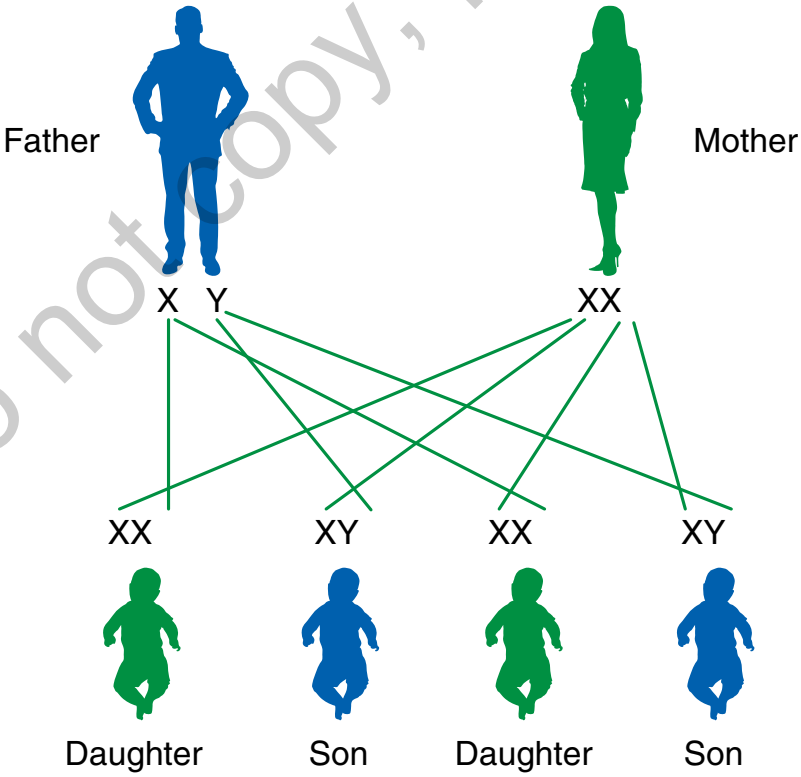
FIGURE 2.2 ■ Sex Determination



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

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differ in appearance, such as hair color, eye color, and height. In about half of fraternal twin pairs, one twin is a boy and the other a girl. DZ twins tend to run in families, suggesting a genetic component that controls the tendency for a woman to release more than one ovum each month (Hazel et al., 2020). Rates of DZ twins also increase with **in vitro fertilization**, maternal age, and with each subsequent birth (Gill et al., 2019; Pison et al., 2015).

Monozygotic (MZ) twins, or identical twins, originate from the same zygote, sharing the same **genotype**, or set of genetic instructions for all physical and psychological characteristics. MZ twins occur when the zygote splits into two distinct separate but identical zygotes that develop into two infants. It is estimated that MZ twins occur in 1 in every 250 births (American College of Obstetricians and Gynecologists, & Society for Maternal-Fetal Medicine, 2014). The causes of MZ twinning are not well understood (McNamara et al., 2016). Rates of MZ twins are not related to maternal age or the number of births, but in vitro fertilization, discussed later, appears to increase the odds of MZ twins (Busnelli et al., 2019; Knopman et al., 2014). DZ twins have become more common in recent decades with advances in reproductive technology, such as in vitro fertilization (Chapter 3) (Rhea et al., 2017).

Patterns of Genetic Inheritance

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

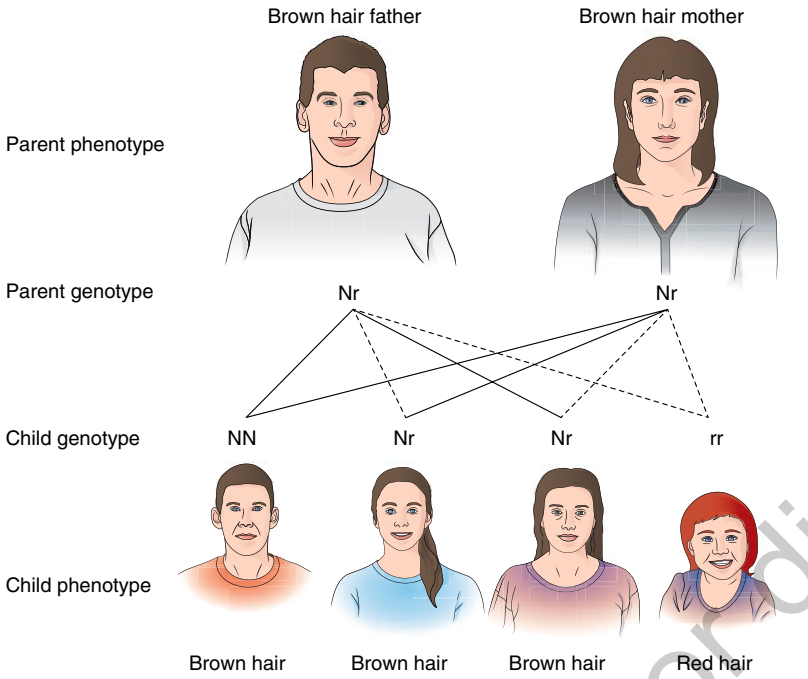
Dominant–Recessive Inheritance

Lynn has red hair while her brother, Jim, does not—and neither do their parents. How did Lynn end up with red hair? These outcomes can be explained by patterns of genetic inheritance, how the sets of genes from each parent interact. As we have discussed, each person has 23 pairs of chromosomes, one pair inherited from the mother and one from the father. The genes within each chromosome can be expressed in different forms, or **alleles**, that influence a variety of physical characteristics.

When alleles of the pair of chromosomes are alike with regard to a specific characteristic, such as hair color, the person is said to be homozygous for the characteristic and will display the inherited trait. If they are different, the person is heterozygous, and the trait expressed will depend on the relations among the genes (Plomin, 2019). 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. Non-red hair is a dominant gene. Other genes, such as for red hair, 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, only the dominant gene is expressed, and the person becomes a carrier of the recessive gene. Both parents have non-red hair but may have homozygous or heterozygous genes for hair color because the gene for non-red hair (symbolized by N in Figure 2.3) is dominant over the gene for red hair (r). If both parents are heterozygous for red hair (Nr), children who inherit a homozygous pair of dominant genes (NN) and others who inherit a heterozygous pair (Nr) will have non-red hair, even though the two genotypes are different. Those who inherit a heterozygous pair (NR) 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), which means that both parents must carry the recessive gene for red hair, even if they display non-red hair.

FIGURE 2.3 ■ Dominant–Recessive Inheritance



Several characteristics are passed through dominant–recessive inheritance (Table 2.1).

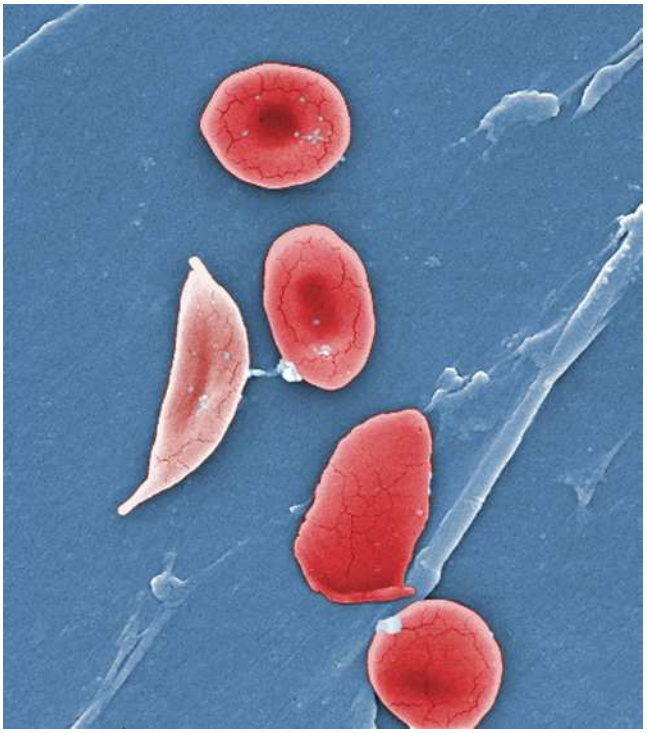
TABLE 2.1 ■ Dominant and Recessive Characteristics

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

Source: McKusick-Nathans Institute of Genetic Medicine (2020).

Incomplete Dominance

In most cases, dominant–recessive inheritance is an oversimplified explanation for patterns of genetic inheritance. **Incomplete dominance** is a genetic inheritance pattern in which both genes jointly influence the characteristic (Knopik et al., 2017). Consider blood type. Neither the alleles for blood type A and B dominate each other. A heterozygous person with the alleles for blood type A and B will express both A and B alleles and have blood type AB.



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.

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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. 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 et al., 2014). Individuals who are homozygous for the recessive sickle cell trait develop sickle cell anemia, a disorder in which 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 and can cause inflammation and damage the blood vessels (Ware et al., 2017). The 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 all of the characteristics of recessive sickle cell alleles, illustrating incomplete dominance. People who carry a single recessive sickle cell gene do not develop full-blown sickle cell anemia (Chakravorty & Williams, 2015). Carriers of the trait for sickle cell anemia tend to function normally but may show some symptoms, such as reduced oxygen distribution throughout the body and exhaustion after exercise (Xu & Thein, 2019).

Polygenic Inheritance

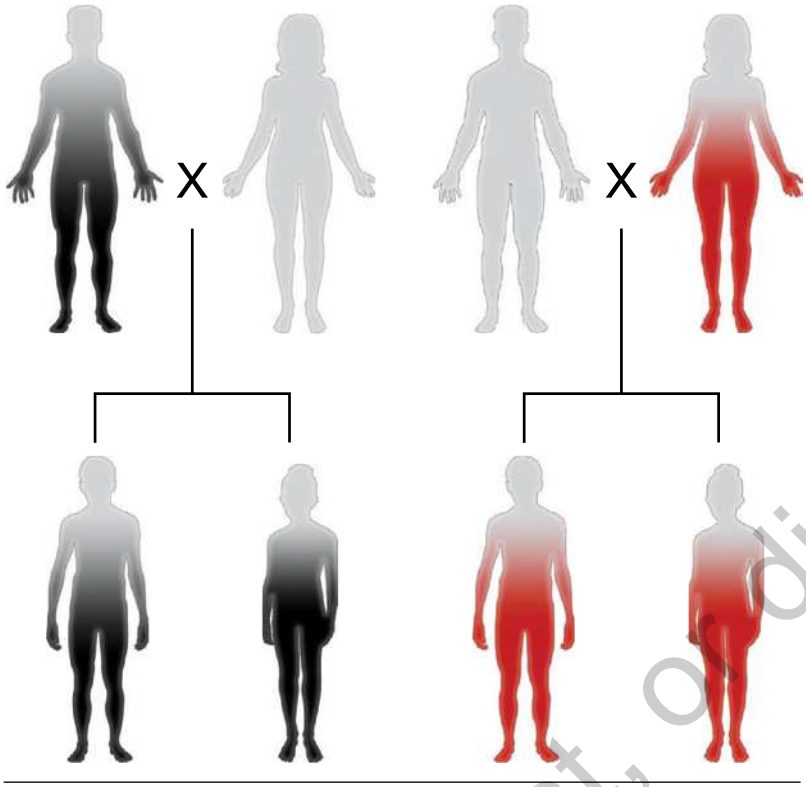
Whereas dominant–recessive and incomplete dominance patterns account for some genotypes, most traits are a function of the interaction of many genes, known as **polygenic inheritance**. Hereditary influences act in complex ways, and researchers cannot trace most characteristics to only one or two genes, but rather, the result of interactions among many genes (Armstrong-Carter et al., 2021). Examples of polygenic traits include height, intelligence, personality, and susceptibility to certain forms of cancer (Bouchard, 2014; Flint et al., 2020; 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.

Genomic Imprinting

The principles of dominant–recessive and incomplete dominance inheritance can account for over 1,000 human traits (Finegold, 2019). 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; Thamban et al., 2020). Consider two conditions that illustrate genomic imprinting: Prader-Willi syndrome and Angelman syndrome. Both syndromes are caused by an abnormality in the 15th chromosome (Kalsner & Chamberlain, 2015). If the individual acquires the chromosome 15 abnormality from the biological father, the individual, whether a daughter or son, will develop Prader-Willi syndrome (Figure 2.4), a set of specific physical and behavioral characteristics including obesity, insatiable hunger, short stature, motor slowness, and mild to moderate developmental delays (Butler et al., 2016).

If the abnormal chromosome 15 arises from the mother, the individual—again, whether it is a daughter or a son—will develop Angelman syndrome, characterized by hyperactivity, thin body frame, seizures, disturbances in gait, severe developmental delay or intellectual disability, and speech

FIGURE 2.4 ■ Genomic Imprinting



Source: Adapted from C. Cristofre Martin (1998).

impairment (Buiting et al., 2016; Dagli & Williams, 2017). Prader-Willi and Angelman syndromes are rare, occurring on average in 1 in 12,000 to 20,000 persons (Kalsner & Chamberlain, 2015; Spruyt et al., 2018). Patterns of genetic inheritance can be complex, yet they follow predictable principles (Table 2.2).

Thinking in Context: Biological Influences

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

TABLE 2.2 ■ Summary: Patterns of Genetic Inheritance

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

CHROMOSOMAL AND GENETIC PROBLEMS

LEARNING OBJECTIVE

2.2 Identify examples of genetic disorders and chromosomal abnormalities.

Many disorders are passed through genetic inheritance, the result of chromosomal abnormalities. Hereditary and chromosomal abnormalities can often be diagnosed prenatally. Others are evident at birth or can be detected soon after an infant begins to develop. Some are discovered only 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 as sickle cell anemia, as well as others that are rare. Some are highly visible and others go unnoticed throughout an individual's life.

Dominant–Recessive Disorders

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. Some diseases are inherited through dominant–recessive patterns (Table 2.3). Few severe disorders are

TABLE 2.3 ■ Diseases Inherited Through Dominant–Recessive Inheritance

Disease	Occurrence	Mode of Inheritance	Description	Treatment
Huntington disease	1 in 20,000	Dominant	Degenerative brain disorder that affects muscular coordination and cognition	No cure; death usually occurs 10 to 20 years after onset
Cystic fibrosis	1 in 2,000–2,500	Recessive	An abnormally thick, sticky mucus clogs the lungs and digestive system, leading to respiratory infections and digestive difficulty	Bronchial drainage, diet, gene replacement therapy
Phenylketonuria (PKU)	1 in 10,000–15,000	Recessive	Inability to digest phenylalanine that, if untreated, results in neurological damage and death	Diet
Sickle cell anemia	1 in 500 African Americans	Recessive	Sickling of red blood cells leads to inefficient distribution of oxygen throughout the body that leads to organ damage and respiratory infections	No cure; blood transfusions, treat infections, bone marrow transplant; death by middle age
Tay-Sachs disease	1 in 3,600 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 (2020).

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 (Ghosh & Tabrizi, 2018; McKusick-Nathans Institute of Genetic Medicine, 2020). 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.

A blood sample to detect PKU is taken from this newborn. Phenylketonuria (PKU) is a genetic disorder in which the body lacks the enzyme that breaks down phenylalanine. Without treatment, the phenylalanine builds up to toxic levels and can damage the central nervous system.

Phenylketonuria (PKU) is a common recessive disorder that prevents the body from producing an enzyme that breaks down phenylalanine, an amino acid, from proteins (McKusick-Nathans Institute of Genetic Medicine, 2020). Without treatment, the phenylalanine builds up quickly to toxic levels that damage the central nervous system, 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 (Camp et al., 2014).

PKU illustrates how genes interact with the environment to produce developmental outcomes. Intellectual disability results from the interaction of the genetic predisposition 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, though they tend to score lower than those without PKU (Hofman et al., 2018; Romani et al., 2017). Some cognitive and psychological problems may appear in childhood and persist into adulthood, particularly difficulty in attention and planning skills, emotional regulation, depression, and anxiety (Christ et al., 2020; Erlich, 2019; Ford et al., 2018; Hawks et al., 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 when young people push boundaries and seek independence (Medford et al., 2017).

X-Linked Disorders

A special instance of the dominant–recessive pattern occurs with genes that are located on the X chromosome (Shah et al., 2017). 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 (Table 2.4). 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.

Fragile X syndrome is an example of a dominant–recessive disorder carried on the X chromosome (Hagerman et al., 2017; Salcedo-Arellano et al., 2020). Because the gene is dominant, it needs to appear on only one X chromosome to be displayed. That means that fragile X syndrome occurs in both males and females. Males with fragile X syndrome typically have a long, narrow face; large ears; and large testes. Fragile X syndrome (FXS) is the most commonly known inherited form of intellectual disability (ID) (Doherty & Scerif, 2017), and children with fragile X syndrome tend to show moderate to severe intellectual disability and problems with executive function (Raspa et al., 2017; Schmitt et al., 2019). Cardiac defects are common as well as several behavioral mannerisms, including poor eye

TABLE 2.4 ■ Diseases Acquired Through X-Linked Inheritance

Syndrome/Disease	Occurrence	Description	Treatment
Color blindness	1 in 12 males	Difficulty distinguishing red from green; less common is difficulty distinguishing blue from green	No cure
Duchenne muscular dystrophy	1 in 3,500 males	Weakness and wasting of limb and trunk muscles; progresses slowly but will affect all voluntary muscles	Physical therapy, exercise, body braces; survival rare beyond late 20s
Fragile X syndrome	1 in 4,000 males and 1 in 8,000 females	Symptoms include cognitive impairment, attention problems, anxiety, unstable mood, long face, large ears, flat feet, and hyperextensible joints, especially fingers	No cure
Hemophilia	1 in 3,000–7,000 males	Blood disorder in which the blood does not clot	Blood transfusions

Source: McKusick-Nathans Institute of Genetic Medicine (2017).

contact and repetitive behaviors such as hand flapping, hand biting, and mimicking others, behaviors common in individuals with autism spectrum disorders (Hagerman et al., 2017; Salcedo-Arellano et al., 2020). Fragile X syndrome is often codiagnosed with autism with estimates of about 40–60% of boys and 16–20% of girls with fragile X syndrome meeting the diagnostic criteria for autism (Bagni & Zukin, 2019; Kaufmann et al., 2017). As carriers, females may show some characteristics of the disorder but tend to display levels of intelligence within the normal or near-normal range.

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 (McKusick-Nathans Institute of Genetic Medicine, 2020; 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 (d'Oiron, 2019). Females, therefore, can carry the gene for hemophilia without exhibiting the disorder. A female carrier has a 50/50 chance of transmitting the gene to each child. 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).

Chromosomal Abnormalities

Down syndrome is the most common cause of intellectual disability. Interventions that encourage children to interact with their physical and social environment can promote motor, social, and emotional development.

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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 et al., 2017; McKusick-Nathans Institute of Genetic Medicine, 2020). 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 often a round face, almond-shaped eyes, and a flattened nose (Antonarakis et al., 2020; Bull, 2020). Children with Down syndrome tend to show delays in physical and motor development relative to other children, and health problems, such as congenital heart defects, vision impairments, poor hearing, and immune system deficiencies (Diamandopoulos & Green, 2018; Morrison & McMahon, 2018; Roizen et al., 2014; Zampieri et al., 2014).

Down syndrome is the most common genetic cause of intellectual developmental disability (Vissers et al., 2016), but children's abilities vary. Generally, individuals with Down syndrome show greater strengths in nonverbal learning and memory relative to their verbal skills (Grieco et al., 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 (Bull, 2020; Næss et al., 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, as compared with about 25 in the 1980s (Glasson et al., 2014; National Association for Down Syndrome, 2020). Many individuals live into their 70s and 80s. However, Down syndrome is associated with premature aging and an accelerated decline of cognitive functioning (Covelli et al., 2016; Ghezzo et al., 2014; Hithersay et al., 2017). Individuals with Down syndrome are at risk to show signs of Alzheimer's disease very early relative to other adults (Antonarakis et al., 2020; Tramutola et al., 2020). This is an example of how disorders and illnesses can be influenced by multiple genes and complex contextual interactions; in this case, Down syndrome and Alzheimer's disease share genetic markers (Handen, 2020; Lee et al., 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 (Table 2.5)

One of the most common sex chromosome abnormalities, with prevalence estimates between 1 in 500 and 1 in 1,000 males, is **Klinefelter syndrome**, in which males are born with an extra X chromosome (XXY) (McKusick-Nathans Institute of Genetic Medicine, 2020; Wistuba et al., 2017). Symptoms range in severity such that some males experience symptoms that impair daily life, but most may be unaware of the disorder until they are tested for infertility (Bird & Hurren, 2016; Gravholt et al., 2018). Severe symptoms include a high-pitched voice, feminine body shape, breast enlargement, and infertility. Many boys and men with Klinefelter syndrome have short stature, a tendency to be overweight, and language and short-term memory impairments that can cause difficulties in learning (Bonomi et al., 2017). As adults, men with Klinefelter syndrome are at risk for a variety of disorders that are more common in women, such as osteoporosis (Juul et al., 2011).

TABLE 2.5 ■ Sex Chromosome Abnormalities

Female Genotype	Syndrome	Description	Prevalence
XO	Turner	Abnormal growth patterns, delayed puberty, lack prominent female secondary sex characteristics, and infertility. Short adult stature, webbing around their neck.	1 in 2,500 females
XXX	Triple X	Grow about an inch or so taller than average with unusually long legs and slender torsos, and show normal development of sexual characteristics and fertility. Because many cases of triple X syndrome often go unnoticed, little is known about the syndrome.	Unknown; many cases go unnoticed
Male Genotype	**Syndrome**	**Description**	**Prevalence**
XXY	Klinefelter	High-pitched voice, short stature, feminine body shape, and infertility. Increased risk for osteoporosis and other disorders that are more common in women.	1 in 1,000 males
XYY	Jacob's syndrome	Accompanied by high levels of testosterone.	Unknown; many cases go unnoticed

A second type of sex chromosome abnormality experienced by men is XYY syndrome, or **Jacob's syndrome**, a condition that causes men to produce high levels of testosterone (McKusick-Nathans Institute of Genetic Medicine, 2017; Pappas et al., 2017). In adolescence, they tend to be slender and show severe acne and poor coordination, but most men with XYY syndrome are unaware that they have a chromosomal abnormality. The prevalence of XYY syndrome is uncertain given that most men go undiagnosed. Females are susceptible to a different set of sex chromosome abnormalities. About 1 in 1,000 females are born with three X chromosomes, known as **triple X syndrome** (McKusick-Nathans Institute of Genetic Medicine, 2020; Wigby et al., 2016). Women with triple X syndrome show an appearance within the norm. They tend to be 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 score lower on inteligence tests or have learning difficulties. Because many cases of triple X syndrome often go unnoticed, little is known about the syndrome.

The sex chromosome abnormality known as **Turner syndrome** occurs when a female is born with only one X chromosome (McKusick-Nathans Institute of Genetic Medicine, 2020). Girls with Turner syndrome show abnormal growth patterns. They show delayed puberty, their ovaries do not develop normally, they do not ovulate, and they are infertile (Culen et al., 2017; Davis et al., 2020). As adults, they are short in stature and often have small jaws with extra folds of skin around their necks (webbing) and lack prominent female secondary sex characteristics such as breasts (Gravholt et al., 2019). Malformations of the heart, diabetes, autoimmune disorders, and early osteoporosis are common (Gravholt et al., 2019). Children with Turner syndrome may show difficulty with visual-spatial reasoning and memory, attention, executive functioning, and motor and math skills (Hutaff-Lee et al., 2019). They are also prone to social difficulties, anxiety, and depression (Christopoulos et al., 2008; Powell & Schulte, 2011). Current estimates put its frequency at 1 in 2,500 worldwide (National Library of Medicine, 2019). If Turner syndrome is diagnosed early, regular injections of human growth hormones can increase stature, and hormones administered at puberty can result in some breast development and menstruation (Culen et al., 2017; Klein et al., 2020).

Mutation

Not all inborn characteristics are inherited. Some result from **mutations**, sudden changes and abnormalities in the structure of genes that occur spontaneously or may be induced by exposure to environmental toxins such as radiation and agricultural chemicals in food. A mutation may involve only one gene or many. It is estimated that as many as one half of all conceptions include mutated chromosomes (Taneri et al., 2020). Most mutations are fatal—the developing organism often dies very soon after conception, often before the woman knows she is pregnant (Sadler, 2018).

Sometimes mutations are beneficial. This is especially true if the mutation is induced by stressors in the environment and provides an adaptive advantage to the individual. The sickle cell gene (discussed earlier in this chapter) is a mutation that originated in areas where malaria is widespread, such as Africa (Ware et al., 2017), and serves a protective role against malaria (Uyoga et al., 2019).

Children who inherited a single sickle cell allele were more resistant to malarial infection and more likely to survive and pass it along to their offspring (Croke et al., 2017; Gong et al., 2013). The sickle cell gene is not helpful in places where malaria is not a risk. The frequency of the gene is decreasing in areas of the world where malaria is uncommon. Only 8% of African Americans are carriers, compared with as many as 30% of Black Africans in some African countries (Maakaron et al., 2012). Therefore, the developmental implications of genotypes—and mutations—are context specific, posing benefits in some contexts and risks in others.

Thinking in Context: Biological Influences

Recall from Chapter 1 that most developmental scientists agree that nature and nuture 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.

Thinking in Context: Lifespan Development

Chromosomal and genetic problems can result in a variety of impairments. How might contextual factors, such as a supportive environment, aid individuals' development? Describe a specific problem or mutation. What environmental conditions might best promote healthy adjustment for individuals with this disorder?

REPRODUCTIVE CHOICES: GENETIC COUNSELING ASSISTED REPRODUCTIVE TECHNOLOGY

LEARNING OBJECTIVE

2.3 Explain the choices of reproductive technology available to individuals and couples who wish to conceive by alternative means.

The likelihood of genetic disorders often can be predicted before conception. Our growing understanding of genetic inheritance has led many couples to consider their own genetic inheritance and what genes they will pass on to their children. Advances in technology permit abnormalities to be detected, and sometimes treated, earlier than ever before.

Genetic Counseling

The popularity of DNA tests has provided many people with information about their genetic makeup. Many prospective parents seek **genetic counseling** to determine the risk that their children will inherit genetic defects and chromosomal abnormalities (Ioannides, 2017). 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 difficulties conceiving or recurrent miscarriages are influenced by chromosomal abnormalities in the male's sperm (Kohn et al., 2016; Softness et al., 2020).

The genetic counselor interviews the couple to construct a family history of heritable disorders for both prospective biological 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, such as Huntington disease, and estimate the likelihood that a child may be affected by a genetic disorder (Nance, 2017). The genetic counselor interprets the results and helps the parents understand genetic concepts by tailoring the explanation to match the parents' knowledge (Abacan et al., 2019).

Parents meet with a genetic counselor to determine the risk that their children will inherit genetic defects and chromosomal abnormalities.

Michelle Del Guercio/Science Source

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 ability to screen

for them, some experts propose 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 et al., 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

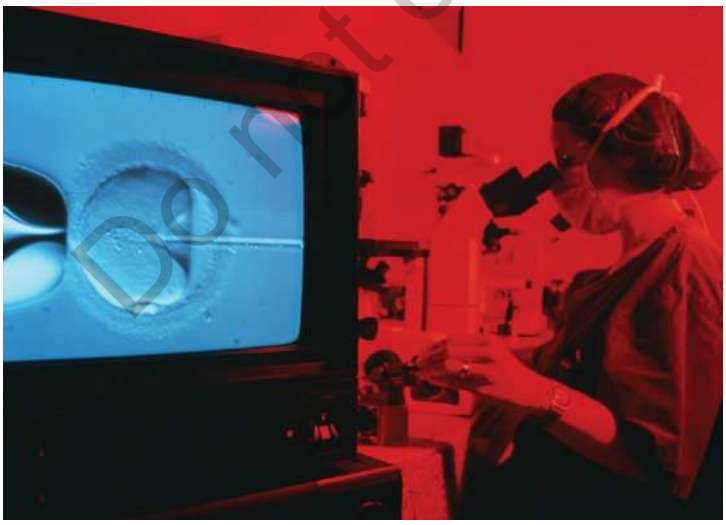
Couples turn to assisted reproductive technology (ART), alternative methods of conception that rely on medical technology, for a variety of reasons. Almost 2% of infants in the United States are conceived through ART (Centers for Disease Control and Prevention, 2020). As noted, some couples at risk for bearing children with genetic or chromosomal abnormalities seek ART. About 15% of couples in the United States experience infertility, the inability to conceive naturally after 1 year of unprotected intercourse (Anwar & Anwar, 2016; Lindahl, 2018). About 35% of the time, factors within the male are identified as contributors to infertility (Centers for Disease Control and Prevention, 2017). In addition, single men and women, as well as gay and lesbian couples, often opt to conceive with the use of ART. There are racial, ethnic, and socioeconomic disparities in the use of ART. White, Asian American, college-educated, and high socioeconomic status women are more likely to give birth with ART than Black and Hispanic women (Janitz et al., 2016; Tierney & Cai, 2019). 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 reproductive technology (Dieke et al., 2017).

Artificial Insemination

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

In Vitro Fertilization

In contrast with artificial insemination, where conception occurs inside of the women's body, in vitro fertilization, introduced in the United States in 1981, initiates conception outside of the woman's body. A woman is prescribed hormones that stimulate the maturation of several ova, which are surgically removed. The ova are placed in a dish and sperm are added. One or more ova are fertilized and the resulting cell begins to divide. After several cell divisions, the cluster of cells is placed in the woman's uterus. If they implant into the uterus and begin to divide, a pregnancy has occurred.



In vitro fertilization is a form of reproductive technology in which an ovum is fertilized outside of the womb.

Mauro Fermariello/Science Source

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

Infants conceived by in vitro fertilization are at higher risk of low birth weight (Centers for Disease Control and Prevention, 2019), although it has been suggested that it is because of maternal factors such as advanced age and health, 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 (Farhi et al., 2019; Fauser et al., 2014). Because in vitro fertilization permits cells to be screened for genetic problems prior to implantation, in vitro infants are not at higher risk of birth defects (Fauser et al., 2014). More than one-third of births from artificial insemination included more than one infant (Sunderam et al., 2019). Multiple gestations increase the risk for low birth weight, prematurity, and other poor outcomes (Sullivan-Pyke et al., 2017).

Surrogacy

Surrogacy is an alternative form of reproduction; a woman (the surrogate) is impregnated and carries a fetus to term and agrees to turn the baby over to a woman, man, or couple who will raise it. Single parents, same-sex couples, and couples in which one or both members are infertile may 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 through surrogacy compared with other methods, including children born to gay father and lesbian mother families (Carone et al., 2018, 2020; Golombok, 2013; Golombok et al., 2017). In addition, mothers of children who were the product of surrogates do not differ from those conceived using other methods and surrogate mothers show no negative effects (Jadva et al., 2015; Söderström-Anttila et al., 2015).

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

Assisted Reproductive Technology and Sex Selection

Gender-reveal parties, in which prospective parents learn about and share the biological sex of their fetus by popping balloons, breaking apart piñatas, lighting fireworks, and other creative means, are popular and signify the relevance of biological sex for many parents. 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 men or women to perform certain kinds of work important to the family or society, or the sex of the couple's other children. Today, many parents cannot only know their child's sex but might choose it. 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 carry.

There are two methods of sex selection: preconception sperm sorting and pre-implantation genetic diagnosis (PGD) (Bhatia, 2018). Preconception sperm sorting involves staining the sperm with a fluorescent dye and then leading them past a laser beam where the difference in DNA content between X- and Y-bearing sperm is visible. PGD creates zygotes within the laboratory by removing eggs from the woman and fertilizing them with sperm. Three days after fertilization a cell from each cluster of cells is extracted to examine the chromosomes and determine whether or not it contains a Y chromosome (i.e., whether it is female or male). The desired male or female embryos are then implanted into the woman's uterus. The second type of sex selection, sperm sorting, entails spinning sperm in a centrifuge to separate those that carry an X or Y chromosome. Sperm with the desired chromosomes are then used to fertilize the ovum either vaginally or through in vitro fertilization.

As sex selection becomes more widely available, parents may seek 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 to avoid transmitting genetic disorders (Bowman-Smart et al., 2020; Robertson & Hickman, 2013). Without the ability to choose a child's sex, some parents might choose not to reproduce (Bowman-Smart et al., 2020). Critics argue that sex selection can lead down a "slippery slope" of selecting for other characteristics—hair color, eye color, intelligence, and more (Dondorp et al., 2013). Moreover, a children's biological sex is different from their gender, the gender-associated behaviors that they adopt (as we will discuss later in this book). (Brown & Stone, 2016). Might children born from gender selection be expected to act in certain sex-typical ways and if they do not, might that disappoint parents?

Others express concerns about societal sex ratio imbalances if sex selection becomes widely practiced (Colls et al., 2009; Robertson & Hickman, 2013). Asian and Eastern European cultures, such as China, South Korea, India, and Azerbaijan, traditionally favor boys over girls (Tafuro & Guilmoto, 2020). Sex ratio imbalances favoring males have occurred in India and China because of female infanticide, neglect and maltreatment, gender-driven abortion, and China's one-child family policy (discussed in Chapter 10) (Bhatia, 2010; Ethics Committee of the American Society for Reproductive Medicine, 2001; Ritchie & Roser, 2019). Thirty-six countries have national laws or policies on sex selection (Mohapatra, 2013). Most prohibit sex selection for nonmedical reasons—Austria, New Zealand, South Korea, Switzerland, Australia, Belgium, Netherlands, India, China, Portugal, Russia, Spain, Turkey, the United Kingdom, and Vietnam ban sex selection (Mohapatra, 2013). The European Union bans socially, nontherapeutically motivated prenatal sex selection (Council of Europe, 1997). The United States does not have a formal policy regarding sex selection (Deeney, 2013). Most fertility clinics offer it (Bayefsky, 2018). Sex selection remains hotly debated in medical journals, with hospital and university ethics boards, and the public.

Thinking in Context: Applied Developmental Science

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. What do you think about parents choosing the sex of their children? In your view, under what conditions is sex selection acceptable?
3. If you were able to choose and selectively reproduce characteristics, apart from sex, what might you choose? Why or why not? What are some practical or ethical issues in selecting a child's characteristics or "designing" a child?

Thinking in Context: Intersectionality

Assisted reproductive technology is not easily available to all individuals and couples. What are some of the barriers to obtaining assisted reproductive technology? Which barriers are most challenging? To what degree is there a need for equity in access to reproductive technology? Why? If so, how might we begin?

REPRODUCTIVE CHOICES: ADOPTION

LEARNING OBJECTIVE

2.4 Compare and contrast characteristics and outcomes of adoption, transracial adoption, and international adoption.

Another reproductive option for prospective parents is **adoption**. Adults, heterosexual and same-sex individuals and couples, have similar motives for adopting children as those who raise biological children (Jennings et al., 2014; Malm & Welti, 2010). They include reasons such as valuing family ties,

continuing a family line, feeling that parenting is a life task, and the desire for a nurturing relationship with a child (Costa & Tasker, 2018; Goldberg et al., 2012).

Adoptive children tend to be raised by parents with higher levels of education and income than other children (Drozd et al., 2018). This is partly due to self-selection and partly because of the screening that adoptive parents must undergo before they can adopt. Adoptive parents have a strong desire and are highly motivated to become parents.

Adoption and Child Outcomes

Overall, adoptive children tend to spend more time with their parents and have more educational resources than other children (Zill, 2015). In kindergarten, they do not differ from nonadopted children in reading and math scores (Tan et al., 2017). Yet many 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 attainment achievement across childhood, adolescence, and emerging adulthood compared with nonadopted comparison groups (Brown et al., 2017; Wiley, 2017).

Adopted children may show more behavior problems and adjustment difficulties than their nonadoptive peers, in some cases persisting into adulthood (Brown et al., 2017; Palacios & Brodzinsky, 2010). For many children, emotional difficulties are transitional, perhaps accounting for mixed findings in outcomes. Some research suggests no differences between adopted and nonadopted children in internalizing problems, such as anxiety and depression (Brown et al., 2019; Wiley, 2017). This is supported by longitudinal research that followed adoptees into middle adulthood and showed few differences in psychological distress; differences were accounted for by differences in childhood family circumstances (Sehmi et al., 2020).

Children's experiences prior to adoption, especially neglect and maltreatment, and their developmental status at the time of adoption influence their short- and long-term adjustment (Balenzano et al., 2018; Finet et al., 2018; Hornfeck et al., 2019). Adopted children tend to experience greater stress prenatally, early in life, prior to adoption, and during the adoption process that influences their long-term adjustment after adoption (Grotevant & McDermott, 2014; Wiley, 2017). The quality of adoptive parent–child relationships influences children's outcomes and the long-term effects of preadoption adversity (Farr & Grotevant, 2019). Children who develop a close bond with adoptive parents tend to show better emotional understanding and regulation, social competence, and also self-esteem (Drozd et al., 2018; Juffer & van IJzendoorn, 2007; Schoemaker et al., 2019). This is true also of children who have experienced emotional neglect, and those effects hold regardless of the age at adoption (Barone et al., 2017).

Transracial Adoption

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). Transracial adoption is associated with academic delays, social and emotional risks such as bullying, racial and ethnic microaggression, and adoption stigma (Branco & Brott, 2018).

Transracial adoptive children, and especially adolescents, may face challenges in ethnic and racial socialization and identity development (Wiley, 2017). Although racial identity development may happen more slowly and entail more challenges for transracially adopted adolescents than nonadopted racial minority adolescents, most develop a firm sense of identity (Hrapczynski & Leslie, 2019). Racial and ethnic socialization is associated with healthy adoptee outcomes, including well-being and positive self-esteem (Montgomery & Jordan, 2018).

Adults choose to adopt for a variety of reasons, such as infertility, the desire to raise a child alone or with a same-sex partner, and to provide a home for a child in need.

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Parents can foster their adoptive children's ethnic and racial socialization by exposing children to their racial and ethnic heritage and provide opportunities for children to learn about and interact with people who identify with their birth race and ethnicity (Hrapczynski & Leslie, 2019). Research with transracially adopted Mexican American adoptees suggests that ethnic identity was associated with living in diverse neighborhoods, parents' awareness of the children's culture, and encouragement for children to learn about and participate in their culture (Montgomery & Jordan, 2018). Close relationships with adoptive parents who engage them with cultural activities are positively associated with academic performance (Montgomery et al., 2020). Transracially adopted children show the most positive self-esteem and well-being outcomes and the least distress when they integrate their racial and cultural heritage with their adoptive culture rather than denying either their adoptive or racial heritage (Mohanty, 2015).

International Adoption

In many countries throughout the world, children without parents are reared in orphanages, often with substandard conditions—without adequate food, clothing, or shelter and with poorly trained caregivers. Such orphanages in countries such as China, Ethiopia, Ukraine, Congo, and Haiti, account 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 handicaps, and growth retardation (The 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. Physical growth stunting is directly associated with the length of institutionalization, but catch-up growth is commonly seen after adoption (Wiley, 2017; Wilson & Weaver, 2009). As with growth, the time spent in an orphanage predicts the degree of developmental delay (Jacobs et al., 2010). Longer institutionalization is associated with delays in 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 adopted children, especially those adopted after the age of 1 (Eigsti et al., 2011). Many 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 (Glennen & Masters, 2002; Mason & Narad, 2005). Virtually all internationally adopted children of all ages show catch-up cognitive growth but many may show long-term deficits in executive function related to the neurological effects of their preadoption experiences, such as deprivation (Canzi et al., 2018; Finet et al., 2019; Merz et al., 2016). The presence of a high-quality parent–child relationship promotes development of language, speech, and academic outcomes (Glennen, 2014; Harwood et al., 2013).

International adoption has become more common in the United States, and there are important challenges that adopted children and families face.

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As adolescents, all children struggle to come to a sense of identity, to figure out who they are. This struggle may be 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 et al., 2015). Like transracially adopted children, internationally adopted children seek to understand their birth

culture and integrate their birth and adopted cultures into their sense of self (Grotevant et al., 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 et al., 2014; Wilson & Weaver, 2009).

Thinking in Context: Intersectionality

1. After many unsuccessful attempts to conceive, a couple in their 40s is considering adopting a child of a different race. What can they expect? What challenges might they encounter?
2. In what ways might the match between the race or ethnicity of the child and parent influence the family's adaptation? Might you expect differences depending on the match between an adoptive parent or child who is Black, white, Latinx, or Asian?
3. Do other factors, such as child's and parent's age, socioeconomic status, or geographic location influence adaptation? Why or why not?
4. What can parents do to support their adopted child of a different race?

Thinking in Context: Lifespan Development

1. In your view, what are the most important challenges internationally adopted infants and their families face? Identify sources and forms of support that might help adopted infants and their parents.
2. At what point in development does an understanding of culture become important in aiding children's adjustment to an adoptive home?
3. As adolescents, many adopted children are driven to learn about their biological origins. How can adoptive parents help their children understand their heritage and adapt to their environment?

PRENATAL DIAGNOSIS

LEARNING OBJECTIVE

2.5 Summarize prenatal diagnostic methods and how genetic disorders may be treated prenatally.

Virtually all pregnant women undergo examinations to determine the health of the fetus. Some women experienced heightened risk for fetal abnormalities. Prenatal testing is recommended when genetic counseling has determined a risk for genetic abnormalities, when the woman is older than age 35, when both parents are members of an ethnicity at risk for particular genetic disorders, or when fetal development appears abnormal (Barlow-Stewart & Saleh, 2012). Technology has advanced rapidly, equipping professionals with an array of tools to assess the health of the fetus.

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

Ultrasound technology provides clear images of the womb, permitting physicians to observe the fetus, measure fetal growth, judge gestational age, reveal the sex of the fetus, detect multiple pregnancies, and determine physical abnormalities in the fetus.

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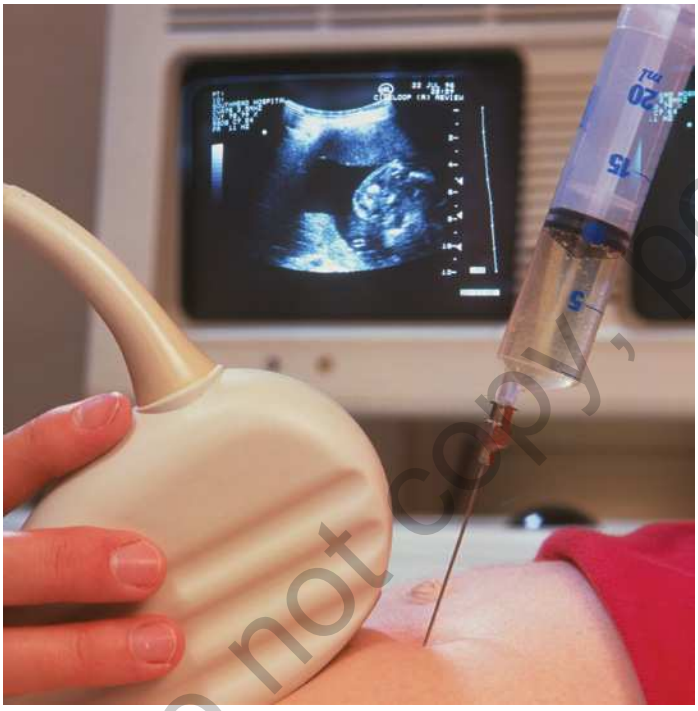

During amniocentesis, ultrasound is used to guide the insertion of a long, hollow needle into the mother's abdomen in order to extract a sample of the amniotic fluid that surrounds the fetus. The amniotic fluid contains fetal cells, which are grown in a laboratory dish and tested for genetic and chromosomal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

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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 or four screenings over the duration of pregnancy are common in order to evaluate fetal development. Repeated ultrasound of the fetus does not appear to affect growth and development (Abramowicz, 2019; Stephenson, 2005).

Fetal MRI applies MRI technology to image the fetus' body and diagnose malformations (Aertsen et al., 2020). It is often used as a follow-up to ultrasound imaging in order 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 (Griffiths et al., 2017; Masselli et al., 2020). MRI in the obstetrical patient is safe for mother and fetus in the second and third trimesters, but it 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 in order 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, but it is safe (Homola & Zimmer, 2019). It is recommended for women aged 35 and over, especially if the woman and partner are both known carriers of genetic diseases (Vink & Quinn, 2018a). Usually amniocentesis is conducted between the 15th and 18th week of pregnancy. Conducted any earlier, an amniocentesis may increase the risk of miscarriage (Akolekar et al., 2015). Test results generally are available about two 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 9 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 one 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 (Salomon et al., 2019; Shim et al., 2014). But 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 (Hartwig et al., 2017; Warsof et al., 2015). Testing can be done after 10 weeks, typically between 10 and 22 weeks. Given that the test involves drawing blood from the mother, there is no risk to the fetus. The use of NIPT has increased dramatically in the United States and other countries (Hui et al., 2017). NIPT can provide accurate sex determination, but NIPT cannot detect as many chromosomal abnormalities as amniocentesis or CVS and is less accurate (Hartwig et al., 2017; Villela et al., 2019). Researchers have identified the entire genome sequence using NIPT, suggesting that someday, NIPT may be as effective as other, more invasive techniques (Tabor et al., 2012). 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. Table 2.6 summarizes methods of prenatal diagnosis.

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 to determine physical abnormalities in the fetus.	Many abnormalities and deformities cannot be easily observed.
Amniocentesis	A small sample of the amniotic fluid that surrounds the fetus is extracted from the mother's uterus through a long, hollow needle inserted into the mother's abdomen. The amniotic fluid contains fetal cells. The fetal cells are grown in a laboratory dish in order to create enough cells for genetic analysis.	It permits a thorough analysis of the fetus's genotype. There is a 100% diagnostic success rate.	Safe, but poses a greater risk to the fetus than ultrasound. If conducted before the 15th week of pregnancy, it may increase the risk of miscarriage.
Chorionic villus sampling (CVS)	Chorionic villus sampling requires studying a small amount of tissue from the chorion, part of the membrane surrounding the fetus, for the presence of chromosomal abnormalities. The tissue sample is obtained through a long needle inserted either abdominally or vaginally, depending on the location of the fetus.	It permits a thorough analysis of the fetus' genotype. CVS is relatively painless, and there is a 100% diagnostic success rate. Can be conducted earlier than amniocentesis, between 10 and 12 weeks.	It may pose a higher rate of spontaneous abortion and limb defects when conducted prior to 10 weeks' gestation.
Fetal MRI	Uses a magnetic scanner to record detailed images of fetal organs and structures.	Provides the most detailed and accurate images available.	It is expensive. At present there is no evidence to suggest that it is harmful to the fetus.
Noninvasive prenatal testing (NIPT)	Cell-free fetal DNA are examined by drawing blood from the mother.	There is no risk to the fetus. It can diagnose several chromosomal abnormalities.	It cannot yet detect the full range of abnormalities. It may be less accurate than other methods. Researchers have identified the entire genome sequence using NIPT, suggesting that someday NIPT may be as effective as other, more invasive techniques.

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

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 utilizes a small camera, inserted through a small incision on the mother's abdomen or cervix and placed into the amniotic sac, which 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 et al., 2014). Surgeons rely on the images provided by fetoscopy to surgically repair defects of the heart, lungs, urinary tract, and other areas (Deprest et al., 2010; Peiro & Scorletti, 2019; Sala et al., 2014).

In addition, researchers believe that one day we may be able to treat many heritable disorders through the use of gene therapy by synthesizing normal genes to replace defective ones. It may someday be possible to sample cells from an embryo, detect harmful genes and replace them with healthy ones, then return the healthy cells to the embryo where they reproduce and correct the genetic defect (Coutelle & Waddington, 2012; Peranteau & Flake, 2020). This approach has been used to correct certain heritable disorders in animals and holds promise for treating humans (Neff, 2019).

Thinking in Context: Applied Developmental Science

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

HEREDITY AND ENVIRONMENT: BEHAVIOR GENETICS

LEARNING OBJECTIVE

2.6 Provide an introduction to the field of behavior genetics, including representative findings.

Our brief introduction to the processes of heredity illustrates the complexity of genetic inheritance. Our genotype, or genetic makeup, inherited from our biological parents, is a biological contributor to all of our observable traits, from hair and eye color to personality, health, and behavior. However, genotypes alone do not determine our **phenotype**, the traits, characteristics, or personalities that we display. Phenotypes result from the interaction of genotypes and our experiences.

Methods of Behavior Genetics

Behavior genetics is the field of study that examines how genes and experience combine to influence the diversity of human traits, abilities, and behaviors (Knopik et al., 2017; Plomin, 2019). Behavior geneticists have discovered that even traits with a strong genetic component, such as height, are modified by environmental influences (Jelenkovic et al., 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 (Briley et al., 2019; Plomin, 2019).

Behavior geneticists seek to estimate the heritability of specific traits and behaviors. **Heritability** refers to the extent to which variation among people on a given characteristic is due to genetic differences. The remaining variation 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 (Fowler-Finn & Boutwell, 2019; Nivard et al., 2017). Behavior 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. Mice can be bred to be very physically active or sedentary by mating highly active mice only with other highly active mice and, similarly, 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 (Schwartz et al., 2018). 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 (Bubac et al., 2020).

For many reasons, especially ethical reasons, people cannot be selectively bred. However, we can observe people who naturally vary in shared genes and environment. Behavior 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 (York, 2020). Twin studies compare identical and fraternal twins to estimate how much of a trait or behavior is attributable to genes. Recall that identical (monozygotic) twins share 100% of their genes because they originated from the same zygote. Like all nontwin siblings, fraternal (dizygotic) twins share 50% of their genes as they resulted from two different fertilized ova and two genetically different zygotes. If genes affect a given attribute, identical twins should be more similar than fraternal twins because identical twins share 100% of their genes, whereas fraternal twins share about half.

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 (York, 2020). If the adopted children share similarities with their biological parents, even though they were not raised by them, it suggests that the similarities are genetic. The similarities are influenced by the environment if the children are more similar to their adoptive parents. Observations of adoptive siblings also shed light on the extent to which attributes and behaviors are influenced by the environment. 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 intelligence scores than do fraternal twins (Plomin, 2019). A classic study of intelligence in over 10,000 twin pairs showed a correlation of .86 for identical and .60 for fraternal twins (Plomin & Spinath, 2004). Table 2.7 summarizes the results of comparisons of intelligence scores from individuals

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

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

Source: Adapted from Bouchard & McGue (1981).

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

who share different genetic relationships with each other. Note that correlations for all levels of kin are higher when they are reared together, supporting the role of environment. Average correlations also rise with increases in shared genes.

Genes contribute to many other traits, such as sociability, temperament, emotionality, and susceptibility to various illnesses such as obesity, heart disease and cancer, anxiety, poor mental health, and a propensity to be physically aggressive (Bralten et al., 2019; Goodarzi, 2018; Morneau-Vaillancourt et al., 2019; Purves et al., 2019; Trucco et al., 2018). Yet even traits that are thought to be heavily influenced by genetics can be modified by physical and social interventions. Growth, body weight, and body height are largely predicted by genetics, yet environmental circumstances and opportunities influence whether genetic potentials are realized (Dubois et al., 2012; Jelenkovic et al., 2016). Even 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.

Thinking in Context: Applied Developmental Science

Imagine that you are a researcher planning to conduct a twin and an adoption study on intelligence, personality, academic achievement, or another topic.

1. What are the advantages and disadvantages of each method?
2. What are some challenges in obtaining participants for these studies?
3. Using the twin approach, how might you determine the genetic and environmental influence on your topic of interest? How does this differ in adoptive studies?
4. What conclusions do you draw about these two types of studies? Which do you prefer and why?

GENE–ENVIRONMENT INTERACTIONS

LEARNING OBJECTIVE

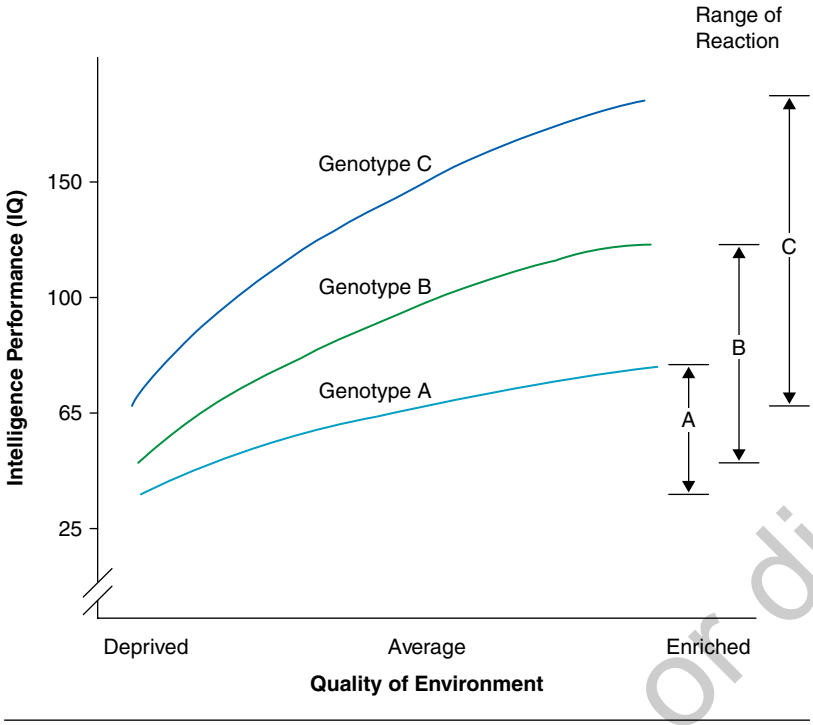
2.7 Describe the interaction of heredity and environment, including gene–environment correlations, gene–environment interactions, and the epigenetic framework.

"You two are so different. Deondre and Denzel, are you sure you're twins?" kidded their aunt. As fraternal twins, Deondre and Denzel share 50% of their genes and are reared in the same home. One might expect them to be quite similar, but their similar genes are not the whole story. Genes and the environment work together in complex ways to determine our characteristics, behavior, development, and health (Morgan et al., 2020; Ritz et al., 2017). **Gene–environment interactions** refer to the dynamic interplay between our genes and our environment. Several principles illustrate these interactions.

Range of Reaction

The effects of the environment depend on the genetic makeup of the individual (Briley et al., 2019). Everyone has a different genetic makeup and therefore responds to the environment in a unique way. In addition, any one genotype can be expressed in a variety of phenotypes. There is a **range of reaction** (see Figure 2.5), a wide range of potential expressions of a genetic trait, depending on environmental opportunities and constraints (Gottlieb, 2007). Consider height. Height is largely a function of genetics, yet an individual may show a range of sizes depending on environment and behavior (Jelenkovic et al., 2016). Consider children born to two very tall parents. They may have the genes to be tall, but unless they have adequate nutrition, they will not fulfill their genetic potential for height. In societies in which nutrition has improved dramatically over a generation, it is common

FIGURE 2.5 ■ Range of Reaction



Source: Adapted from Gottlieb (2007).

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; Morgan et al., 2020). In this way, genetics sets the range of development outcomes and the environment influences where, within the range, that person will fall. However, gene–environment interactions are complex and often difficult to predict, partly because individuals vary in their sensitivity to environmental stimuli (Belsky & Hartman, 2014). Some children may be more affected by environmental stimuli due to their genetic makeup (Briley et al., 2019).

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; Takahashi, 2019). Infants follow an age-related sequence of motor development, from crawling, to walking, to running. Around the world, most infants walk at about 12 months of age. Generally, only extreme experiences or changes in the environment can prevent this developmental sequence from occurring (Adolph & Franchak, 2017). Children reared in impoverished international orphanages and exposed to extreme environmental deprivation demonstrated delayed motor development, with infants walking 5months to a year later than expected (Miller et al., 2008; Chaibal et al., 2016).

Motor development is not entirely canalized because some minor changes in the environment can subtly alter its pace and timing. Practice facilitates stepping movements in young infants, prevents the disappearance of stepping movements in the early months of life, and leads to an earlier onset of walking (Adolph & Hoch, 2019). These observations demonstrate that even highly canalized traits, such as motor development, which largely unfolds via maturation, can be subtly influenced by contextual factors.

Gene–Environment Correlations

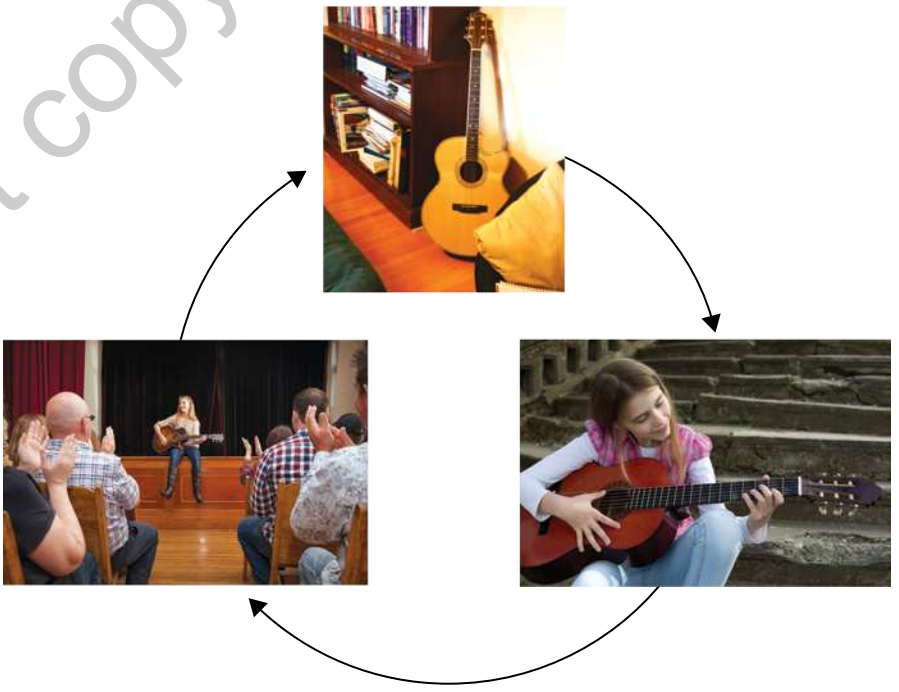
Heredity and environment are each powerful influences on development. Not only do they interact, but environmental factors often support hereditary traits (Briley et al., 2019; Saltz, 2019; Scarr & McCartney, 1983). **Gene–environment correlation** refers to the finding that many genetically influenced traits tend to be associated with environmental factors that promote their development (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, reactive, 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 they also correspond to their child's genotype—an example of a passive gene–environment correlation (Wilkinson et al., 2013). It is a passive gene–environment correlation because it occurs regardless of the child's behavior. Parents might provide genes that predispose a child to develop music ability and create a home environment that supports the development of music ability, such as by playing music in the home and owning musical instruments (Corrigall & Schellenberg, 2015) (Figure 2.6). This type of gene–environment correlation tends to occur early in life because parents create rearing environments for their infants and young children.

People naturally evoke responses from others and the environment, just as the environment and the actions of others evoke responses from the individual. In an evocative gene–environment correlation, a child's genetic traits (e.g., personality characteristics including openness to experience) influence the social and physical environment, which shape development in ways that support the genetic trait (Pieters et al., 2015; Saltz, 2019). Active, happy infants tend to receive more adult attention than do passive or moody infants (Deater-Deckard & O'Connor, 2000), and even among infant twins reared in the same family, the more outgoing and happy twin receives more positive attention than does the more subdued twin (Deater-Deckard, 2001). Why? Babies who are cheerful and smile often influence their social world by evoking smiles and affection from others, including their parents, which in turn

FIGURE 2.6 ■ Gene–Environmental Correlation

The availability of instruments in the home corresponds to the child's musical abilities, and they begin to play guitar (passive gene–environment correlation). As they play guitar, they evoke positive responses in others, increasing their interest in music (evocative gene–environment correlation). Over time, they seek opportunities to play, such as performing in front of an audience (niche-picking).



Sources: iStock/Signature; iStock/Essentials; iStock/Essentials

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

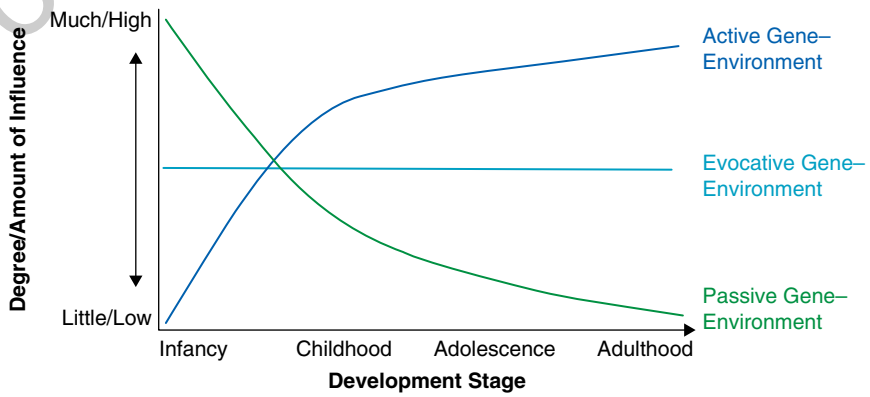
Children also take a hands-on role in shaping their development. Recall from Chapter 1 that a major theme in understanding human development is the finding that individuals are active in their development; here we have an example of this theme. As children grow older, they have increasing freedom in 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. The child with a genetic trait for interest and ability in music actively seeks experiences and environments that support that trait, such as friends with similar interests and after-school music classes (Corrigall & Schellenberg, 2015). This tendency to actively seek out experiences and environments compatible and supportive of our genetic tendencies is called **niche-picking** (Saltz, 2019; Scarr & McCartney, 1983).

The strength of passive, evocative, and active gene–environment correlations changes with development, as shown in Figure 2.7 (Lynch, 2016; Scarr, 1992). Passive gene–environment correlations are common at birth as caregivers determine infants' experiences. Correlations between their genotype and environment tend to occur because their environments are made by genetically similar parents (Armstrong-Carter et al., 2021). 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 & Von Stumm, 2018; York, 2020).

Gene–Environment (G x E) Interactions

We have seen that behavior is influenced by gene–environment interactions. Genes may provide a reaction range through which environmental factors act. Some genes severely limit developmental outcomes (canalization). Although behavior geneticists have learned a great deal about genetic influences on behavior, effects are often unpredictable (Flint et al., 2020). The effects of genes vary with environmental influences and not all genotypes respond to environmental influences in the same way (Fowler-Finn & Boutwell, 2019).

FIGURE 2.7 ■ Development Stage and Gene–Environment Correlations



In a classic study, Caspi et al. (2002) followed a sample of boys from birth until adulthood. Although children who experience child maltreatment, or abuse, tend to show developmental and behavioral problems, the effects of maltreatment varied among the boys. Upon further study, the researchers were surprised to find that the link between maltreatment and violence varied with the gene that controls monoamine oxidase A (MAOA), an enzyme that regulates specific chemicals in the brain. Only boys who carried a certain form of this gene were at risk for becoming violent after experiencing maltreatment. Specifically, there are two versions of the gene that controls MAOA: one produces high levels of the enzyme, and the other produces low levels.

Boys who experienced abuse and other traumatic experiences were about twice as likely to develop problems with aggression, violence, and to even be convicted of a violent crime—but only if they carried the low-MAOA gene. Maltreated boys who carried the high-MAOA gene were no more likely to become violent than nonmaltreated boys. In addition, the presence of the low-MAOA gene itself was not associated with violence. The low-MAOA gene predicted violence only for boys who experience abuse early in life. These findings have been replicated in another 30-year longitudinal study of boys (Fergusson et al., 2011) as well as a meta-analysis of 27 studies (Byrd & Manuck, 2014).

Similar findings of an MAOA gene × environment interaction in which low MAOA, but not high MAOA, predicts negative outcomes in response to childhood adversity has been extended to include other mental health outcomes such as antisocial personality disorder and depression (Beach et al., 2010; Cicchetti et al., 2007; Manuck & McCaffery, 2014; Nikulina et al., 2012). Many of these studies have examined only males. Females show a more mixed pattern, with some studies showing that girls display the MAOA gene × environment interaction on emotional reactivity and aggression but to a much lesser extent than boys, whereas other studies suggest no relationship (Byrd & Manuck, 2014; Byrd et al., 2018). The MAOA gene illustrates gene–environment interactions, but gene–environment interactions determine the effects of many genes. The 5-HTTLPR gene, responsible for regulating specific chemicals in the brain, interacts with environmental factors to influence parenting sensitivity, depression, stress, and responses to trauma (Baião et al., 2020; Li et al., 2013).

We have seen that gene–environment interactions influence development and behavior. Genes may provide a reaction range through which environmental factors act. Some genes severely limit developmental outcomes (canalization). Although behavior geneticists have learned a great deal about genetic influences on behavior, their effects are often unpredictable (Flint et al., 2020). Just as some genes might increase our susceptibility to environmental risks, others might increase our sensitivity to, and the effectiveness of, environmental interventions (Bakermans-Kranenburg & van IJzendoorn, 2015; Chhangur et al., 2017). The effects of genes vary with environmental influences and not all genotypes respond to environmental influences in the same way (Fowler-Finn & Boutwell, 2019). Conclusions about gene–environment interactions pertain to populations, not individuals. Therefore, findings from behavior genetic research cannot predict individual behavior (Turkheimer, 2019). A final important criticism of behavior genetic research is that, like many other areas of research, its samples are not diverse. Ethnically diverse samples and those of low socioeconomic status are underrepresented, limiting conclusions (Sirugo et al., 2019).

Not all children exposed to adversity experience negative outcomes. Genes, such as MAOA, influence children's sensitivity to maltreatment.

FatCamera/Getty Images

Epigenetic Framework

Development is the product of a dynamic interaction of biological and contextual forces. Genes provide a blueprint for development, but phenotypic outcomes, individuals' characteristics, are not predetermined but vary with environmental factors. Recently, scientists have determined that environmental factors do not simply interact with genes to determine people's traits, but they can determine how genes are expressed through a process

FIGURE 2.8 ■ Altering the Epigenome

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

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

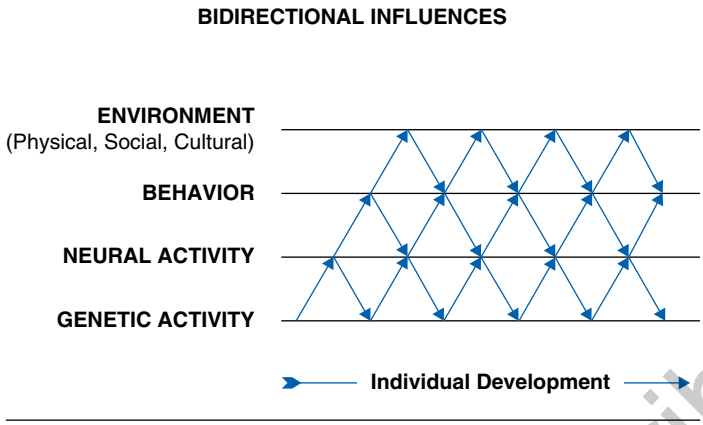
known as **epigenetics** (Carlberg & Molnar, 2019; Moore, 2017). The term epigenetics literally means "above the gene." The epigenome is a molecule that stretches along the length of DNA and provides instructions to genes, determining how they are expressed—whether they are turned on or off. The epigenome carries the instructions that determine what each cell in your body will become, whether heart cell, muscle cell, or brain cell. Those instructions are carried out by turning genes on and off.

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

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. A groundbreaking study showed that yellow agouti mice can produce offspring that look very different (Waterland & Jirtle, 2003). The mice in Figure 2.8 both carry the agouti gene, yet they look very different; the brown mouse is slender, lean, and has a low risk of developing diabetes and cancer, living well into old age. Why are these mice so different? Epigenetics. In the case of the yellow and brown mice, the phenotype of the brown mice has been altered, but the DNA remains the same. Both carry the agouti gene, but in the yellow mouse, the agouti gene is turned on all the time. In the brown mouse, it is turned off.

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

FIGURE 2.9 ■ Epigenetic Framework



Source: Gottlieb (2007).

Epigenetic processes also influence human development. Consider brain development (O'Donnell & Meaney, 2020). Providing infants with a healthy diet and opportunities to explore the world will support the development of brain cells, governed by epigenetic mechanisms that switch genes on and off. Conversely, epigenetic changes that accompany exposure to toxins or extreme trauma might suppress the activity of some genes, potentially negatively influencing brain development. In this way, individuals' neurological capacities are the result of epigenetic interactions among genes and contextual factors (Lerner & Overton, 2017) (Figure 2.9). Interactions between heredity and environment change throughout development as does the role we play in constructing environments that support our genotypes, influence our epigenome, and determine who we become (Lickliter & Witherington, 2017).

Perhaps the most surprising finding emerging from animal studies of epigenetics 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 (Legoff et al., 2019; Szyf, 2015). This means that what you eat and do today could affect the epigenome—the development, characteristics, and health—of your children, grandchildren, and great-grandchildren (Bošković & Rando, 2018; Grover & Jenkins, 2020; Vanhees et al., 2014).

Thinking in Context: Lifespan Development

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

Thinking in Context: Biological Influences

1. Considering the research on epigenetics, what can you do to protect your epigenome? What kinds of behavioral and contextual factors might influence your epigenome?
2. If some genes may be protective in particular contexts, should scientists learn how to turn them on? Should scientists learn to turn off genes that might increase risks in particular contexts? Why or why not?

APPLY YOUR KNOWLEDGE

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

Many years later, Jenna was surprised to learn that she had a twin sister, Tasha. Separated at birth, Jenna and Tasha became aware of each other in their early 40s. Jenna was stunned yet couldn't wait to meet her twin sister. Upon meeting, Jenna and Tasha were surprised to find that they were not exactly the same. Whereas Jenna was athletic and lithe, Tasha was more sedentary and substantially heavier than Jenna. Unlike Jenna, Tasha grew up in a home far from the beach and with little access to outdoor activities. Instead, Tasha's interest was writing. As a child, she'd write stories and share them with others. She sought out opportunities to write and chose a college with an exceptional writing program. Both Jenna and Tasha excelled in college, as they did throughout their education, and earned nearly identical scores on the SAT.

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

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

CHAPTER SUMMARY

2.1 Discuss the genetic foundations of development.

Genes are composed of stretches of deoxyribonucleic acid (DNA). Most cells in the human body reproduce through mitosis, but sex cells reproduce by meiosis, creating gametes with 23 single, unpaired chromosomes. Some genes are passed through dominant–recessive inheritance, in which some genes are dominant and will always be expressed, and others are recessive and will only be expressed if paired with another recessive gene. Other patterns include incomplete dominance and genomic imprinting. Most traits are polygenic, the result of interactions among many genes.

2.2 Identify examples of genetic disorders and chromosomal abnormalities.

Genetic disorders carried through dominant–recessive inheritance include PKU, 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. Fragile X syndrome is an example of a dominant–recessive disorder carried on the X chromosome. Other X-linked genetic disorders include Klinefelter syndrome, Jacob's syndrome, triple X syndrome, and Turner syndrome. Some disorders, such as trisomy 21, known as Down syndrome, are the result of chromosomal abnormalities. Others result from mutations.

2.3 Explain the choices of reproductive technology available to individuals and couples who wish to conceive by alternative means.

Individuals and couples turn to assisted reproductive technology (ART) for a variety of reasons, including infertility, to reduce the risk of genetic or chromosomal abnormalities, or to conceive without a partner. Artificial insemination, the simplest ART, involves injecting sperm into a woman. In vitro fertilization involves fertilizing ova with sperm in a dish and implanting the resulting cluster of cells in the woman's uterus. 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 it.

2.4 Compare and contrast characteristics and outcomes of adoption, transracial adoption, and international adoption.

Adoptive children tend to be raised by parents with higher levels of education and have more educational resources than other children. Yet adoption is associated with lower academic attainment achievement and sometimes transient behavior problems. For internationally adopted children, the time spent in an orphanage predicts the degree of developmental delay, but virtually all internationally adopted children show some catch-up cognitive growth. For both transracial and internationally adopted children, racial and ethnic socialization is associated with healthy outcomes, including well-being, positive self-esteem, and identity. Children's experiences prior to adoption, especially neglect and maltreatment, and their developmental status at the time of adoption, influence their short- and long-term adjustment. Generally, adopted children overall show great developmental gains and resilience in physical, cognitive, and emotional development.

2.5 Summarize prenatal diagnostic methods and how genetic disorders may be treated prenatally.

Ultrasound enables physicians to observe the fetus, measure fetal growth, judge gestational age, and determine physical abnormalities in the fetus. Fetal MRI applies MRI technology to image the fetus' body and diagnose malformations, and it is often used as a follow-up to ultrasound imaging. Amniocentesis involves extracting a small sample of the amniotic fluid that surrounds the fetus. The extracted fetal cells are grown in a laboratory dish and then analyzed. Chorionic villus sampling (CVS) also samples genetic material and can be conducted earlier than amniocentesis. Noninvasive prenatal testing (NIPT) screens the mother's blood to detect chromosomal abnormalities, but it is not as accurate as amniocentesis and CVS. Fetoscopy involves inserting a camera into the womb to examine and perform procedures, including surgery, on the fetus during pregnancy.

2.6 Provide an introduction to the field of behavior genetics, including representative findings.

Behavior 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.

2.7 Describe the interaction of heredity and environment, including gene–environment correlations, gene–environment interactions, and the epigenetic framework.

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. People's genes and environment interact in complex ways such that the effects of experience may vary with a person's genes. The epigenetic framework is a model for understanding the dynamic, ongoing interactions between heredity and environment whereby the epigenome's instructions to turn genes on and off throughout development are influenced by the environment.

KEY TERMS

Adoption (p. 66)
Alleles (p. 54)
Amniocentesis (p. 70)
Artificial insemination (p. 64)
Behavior genetics (p. 72)
Canalization (p. 75)
Chorionic villus sampling (CVS) (p. 70)
Chromosomes (p. 52)
Dizygotic twins (p. 81)
DNA (deoxyribonucleic acid) (p. 44)
Dominant–recessive inheritance (p. 54)
Down syndrome (p. 60)
Epigenetics (p. 79)
Fetal MRI (p. 70)
Fetoscopy (p. 72)
Fragile X syndrome (p. 59)
Gametes (p. 52)
Gene–environment correlation (p. 76)
Gene–environment interactions (p. 74)
Genes (p. 52)
Genetic counseling (p. 63)
Genomic imprinting (p. 56)
Genotype (p. 54)
Hemophilia (p. 60)
Heritability (p. 72)
Incomplete dominance (p. 55)
In vitro fertilization (p. 54)
Jacob's syndrome (p. 62)
Klinefelter syndrome (p. 61)
Meiosis (p. 52)
Mitosis (p. 52)
Monozygotic twins (p. 62)
Mutations (p. 62)
Niche-picking (p. 77)
Noninvasive prenatal testing (p. 71)
Phenotype (p. 72)
Phenylketonuria (PKU) (p. 59)
Polygenic inheritance (p. 56)
Range of reaction (p. 74)
Sickle cell trait (p. 56)
Surrogacy (p. 65)
Triple X syndrome (p. 62)
Turner syndrome (p. 62)
Ultrasound (p. 69)
Zygote (p. 52)