Learning Objectives

By the end of this section, you will be able to:

- Explain the basic principles of the theory of evolution by natural selection
- Describe the differences between genotype and phenotype
- Discuss how gene-environment interactions are critical for expression of physical and psychological characteristics

Psychological researchers study genetics in order to better understand the biological factors that contribute to certain behaviors. While all humans share certain biological mechanisms, we are each unique. And while our bodies have many of the same parts—brains and hormones and cells with genetic codes—these are expressed in a wide variety of behaviors, thoughts, and reactions.

Why do two people infected by the same disease have different outcomes: one surviving and one succumbing to the ailment? How are genetic diseases passed through family lines? Are there genetic components to psychological disorders, such as depression or schizophrenia? To what extent might there be a psychological basis to health conditions such as childhood obesity?

To explore these questions, let's start by focusing on a specific genetic disorder, sickle cell anemia, and how it might manifest in two affected sisters. Sickle-cell anemia is a genetic condition in which red blood cells, which are normally round, take on a crescent-like shape (Figure 3.2). The changed shape of these cells affects how they function: sickle-shaped cells can clog blood vessels and block blood flow, leading to high fever, severe pain, swelling, and tissue damage.

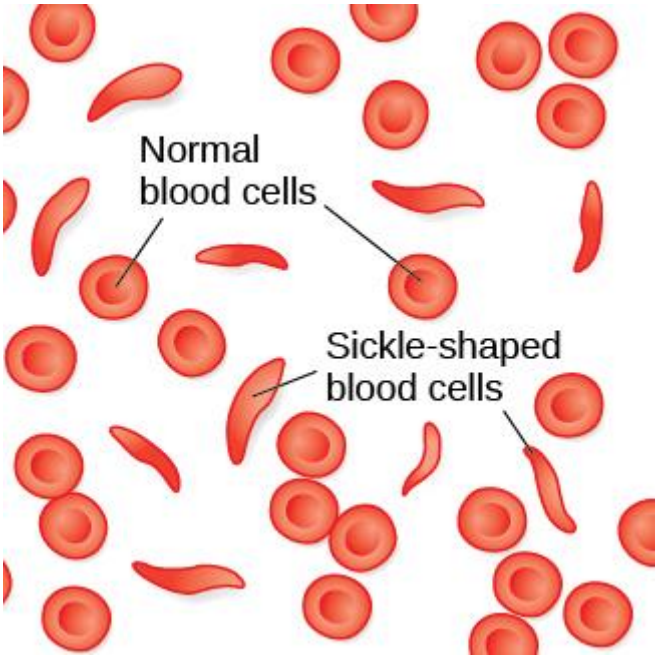


Figure 3.2 Normal blood cells travel freely through the blood vessels, while sickle-shaped cells form blockages preventing blood flow.

Many people with sickle-cell anemia—and the particular genetic mutation that causes it—die at an early age. While the notion of “survival of the fittest” may suggest that people with this disorder have a low survival rate and therefore the disorder will become less common, this is not the case. Despite the negative evolutionary effects associated with this genetic mutation, the sickle-cell gene remains relatively common among people of African descent. Why is this? The explanation is illustrated with the following scenario.

Imagine two young women—Luwi and Sena—sisters in rural Zambia, Africa. Luwi carries the gene for sickle-cell anemia; Sena does not carry the gene. Sickle-cell carriers have one copy of the sickle-cell gene but do not have full-blown sickle-cell anemia. They experience symptoms only if they are severely dehydrated or are deprived of oxygen (as in mountain climbing). Carriers are thought to be immune to malaria (an often deadly disease that is widespread in tropical climates) because changes in their blood chemistry and immune functioning prevent the malaria parasite from having its effects (Gong, Parikh, Rosenthal, & Greenhouse, 2013). However, full-blown sickle-cell anemia, with two copies of the sickle-cell gene, does not provide immunity to malaria.

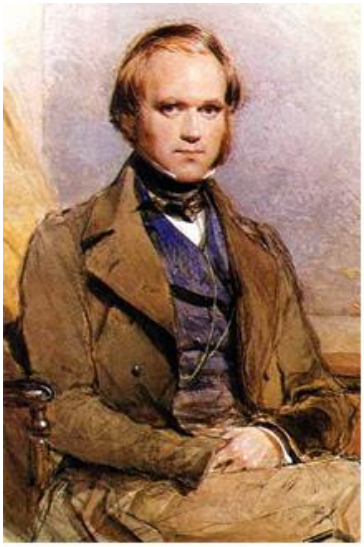
While walking home from school, both sisters are bitten by mosquitoes carrying the malaria parasite. Luwi is protected against malaria because she carries the sickle-cell mutation. Sena, on the other hand, develops malaria and dies just two weeks later. Luwi survives and eventually has children, to whom she may pass on the sickle-cell mutation.

LINK TO LEARNING

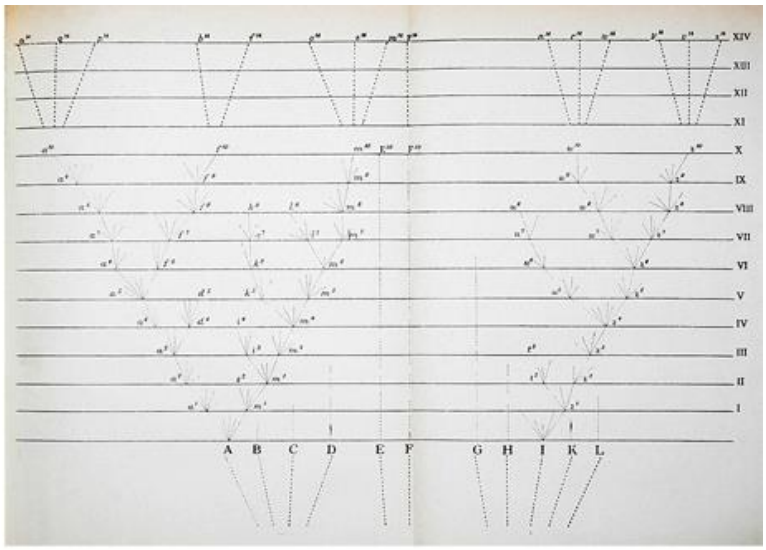
Visit this website about how a mutation in DNA leads to sickle cell anemia to learn more.

Malaria is rare in the United States, so the sickle-cell gene benefits nobody: the gene manifests primarily in minor health problems for carriers with one copy, or a severe full-blown disease with no health benefits for carriers with two copies. However, the situation is quite different in other parts of the world. In parts of Africa where malaria is prevalent, having the sickle-cell mutation does provide health benefits for carriers (protection from malaria).

The story of malaria fits with Charles Darwin's **theory of evolution by natural selection** (Figure 3.3). In simple terms, the theory states that organisms that are better suited for their environment will survive and reproduce, while those that are poorly suited for their environment will die off. In our example, we can see that, as a carrier, Luwi’s mutation is highly adaptive in her African homeland; however, if she resided in the United States (where malaria is rare), her mutation could prove costly—with a high probability of the disease in her descendants and minor health problems of her own.



(a)



(b)

Figure 3.3 (a) In 1859, Charles Darwin proposed his theory of evolution by natural selection in his book, *On the Origin of Species*. (b) The book contains just one illustration: this diagram that shows how species evolve over time through natural selection.

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Two Perspectives on Genetics and Behavior

It's easy to get confused about two fields that study the interaction of genes and the environment, such as the fields of evolutionary psychology and behavioral genetics. How can we tell them apart?

In both fields, it is understood that genes not only code for particular traits, but also contribute to certain patterns of cognition and behavior. Evolutionary psychology focuses on how universal patterns of behavior and cognitive processes have evolved over time. Therefore, variations in cognition and behavior would make individuals more or less successful in reproducing and passing those genes on to their offspring. Evolutionary psychologists study a variety of psychological phenomena that may have evolved as adaptations, including fear response, food preferences, mate selection, and cooperative behaviors (Confer et al., 2010).

Whereas evolutionary psychologists focus on universal patterns that evolved over millions of years, behavioral geneticists study how individual differences arise, in the present, through the interaction of genes and the environment. When studying human behavior, behavioral geneticists often employ twin and adoption studies to research questions of interest. Twin studies compare the likelihood that a given behavioral trait is shared among identical and fraternal twins; adoption studies compare those rates among biologically related relatives and adopted relatives. Both approaches provide some insight into the relative importance of genes and environment for the expression of a given trait.

LINK TO LEARNING

Watch this interview with renowned evolutionary psychologist David Buss to learn more about how a psychologist approaches evolution and how this approach fits within the social sciences.

Genetic Variation

Genetic variation, the genetic difference between individuals, is what contributes to a species' adaptation to its environment. In humans, genetic variation begins with an egg, about 100 million sperm, and fertilization. Roughly once per month, active ovaries release an egg from follicles. During the egg's journey from the ovary through the fallopian tubes, to the uterus, a sperm may fertilize the egg.

The egg and the sperm each contain 23 chromosomes. **Chromosomes** are long strings of genetic material known as **deoxyribonucleic acid (DNA)**. DNA is a helix-shaped molecule made up of nucleotide base pairs. In each chromosome, sequences of DNA make up **genes** that control or partially control a number of visible characteristics, known as traits, such as eye color, hair color, and so on. A single gene may have multiple possible variations, or alleles. An **allele** is a specific version of a gene. So, a given gene may code for the trait of hair color, and the different alleles of that gene affect which hair color an individual has.

When a sperm and egg fuse, their 23 chromosomes combine to create a zygote with 46 chromosomes (23 pairs). Therefore, each parent contributes half the genetic information carried by the offspring; the resulting physical characteristics of the offspring (called the phenotype) are determined by the interaction of genetic material supplied by the sperm and egg (called the genotype). A person's **genotype** is the genetic makeup of that individual. **Phenotype**, on the other hand, refers to the individual's inherited physical characteristics, which are a combination of genetic and environmental influences (Figure 3.4).

(a) (b)

Figure 3.4 (a) Genotype refers to the genetic makeup of an individual based on the genetic material (DNA) inherited from one's genetic contributors. (b) Phenotype describes an individual's observable characteristics, such as hair color, skin color, height, and build. (credit a: modification of work by Caroline Davis; credit b: modification of work by Cory Zanker)

Note that, in genetics and reproduction, "parent" is often used to describe the individual organisms that contribute genetic material to offspring, usually in the form of gamete cells (sperm and egg). The concept of a genetic parent is distinct from social and legal concepts of parenthood, and may differ from those whom people consider their parents.

Most traits are controlled by multiple genes, but some traits are controlled by one gene. A characteristic like cleft chin, for example, is influenced by a single gene from each parent. In this example, we will call the gene for cleft chin "B," and the gene for smooth chin "b." Cleft chin is a dominant trait, which means that having the **dominant allele** either from one parent (Bb) or both parents (BB) will always result in the phenotype associated with the dominant allele. When someone has two copies of the same allele, they are said to be **homozygous** for that allele. When someone has a combination of alleles for a given gene, they are said to be **heterozygous**. For example, smooth chin is a recessive trait, which means that an individual will only display the smooth chin phenotype if they are homozygous for that **recessive allele** (bb).

Imagine that a person with a cleft chin mates with a person with a smooth chin. What type of chin will their offspring have? The answer to that depends on which alleles each parent carries. If the person with a cleft is homozygous for cleft chin (BB), their offspring will always have cleft chin. It gets a little more complicated, however, if the person is heterozygous for this gene (Bb). Since the other person has a smooth chin—therefore homozygous for the recessive allele (bb)—we can expect the offspring to have a 50% chance of having a cleft chin and a 50% chance of having a smooth chin (Figure 3.5).

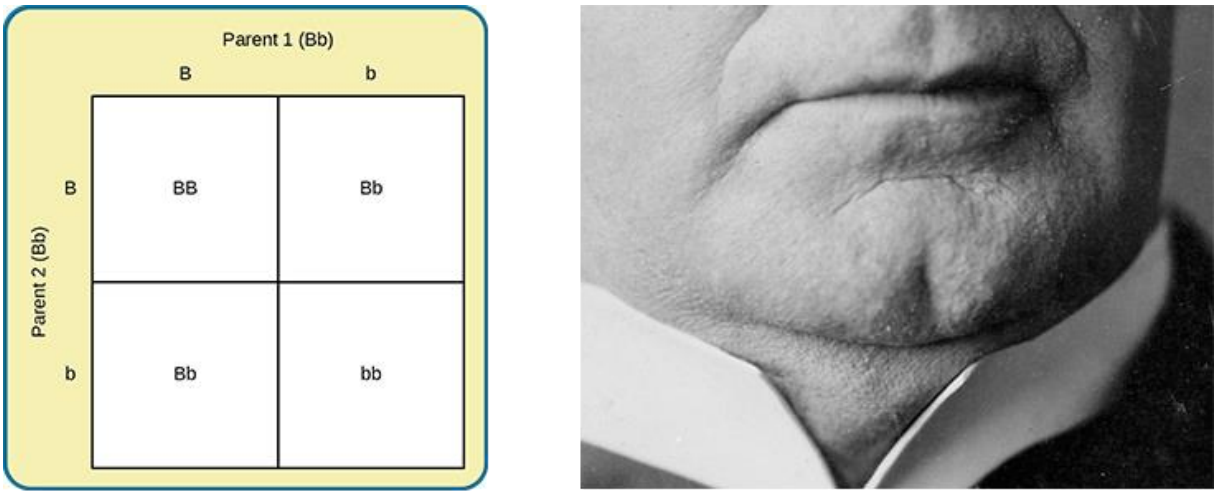

Figure 3.5 (a) A Punnett square is a tool used to predict how genes will interact in the production of offspring. The capital B represents the dominant allele, and the lowercase b represents the recessive allele. In the example of the cleft chin, where B is cleft chin (dominant allele), wherever a pair contains the dominant allele, B, you can expect a cleft chin phenotype. You can expect a smooth chin phenotype only when there are two copies of the recessive allele, bb. (b) A cleft chin, shown here, is an inherited trait.

In sickle cell anemia, heterozygous carriers (like Luwi from the example) can develop blood resistance to malaria infection while those who are homozygous (like Sena) have a potentially lethal blood disorder. Sickle-cell anemia is just one of many genetic disorders caused by the pairing of two recessive genes. For example, phenylketonuria (PKU) is a condition in which individuals lack an enzyme that normally converts harmful amino acids into harmless byproducts. If someone with this condition goes untreated, they will experience significant deficits in cognitive function, seizures, and an increased risk of various psychiatric disorders. Because PKU is a recessive trait, each parent must have at least one copy of the recessive allele in order to produce a child with the condition (Figure 3.6).

So far, we have discussed traits that involve just one gene, but few human characteristics are controlled by a single gene. Most traits are **polygenic**: controlled by more than one gene. Height is one example of a polygenic trait, as are skin color and weight.

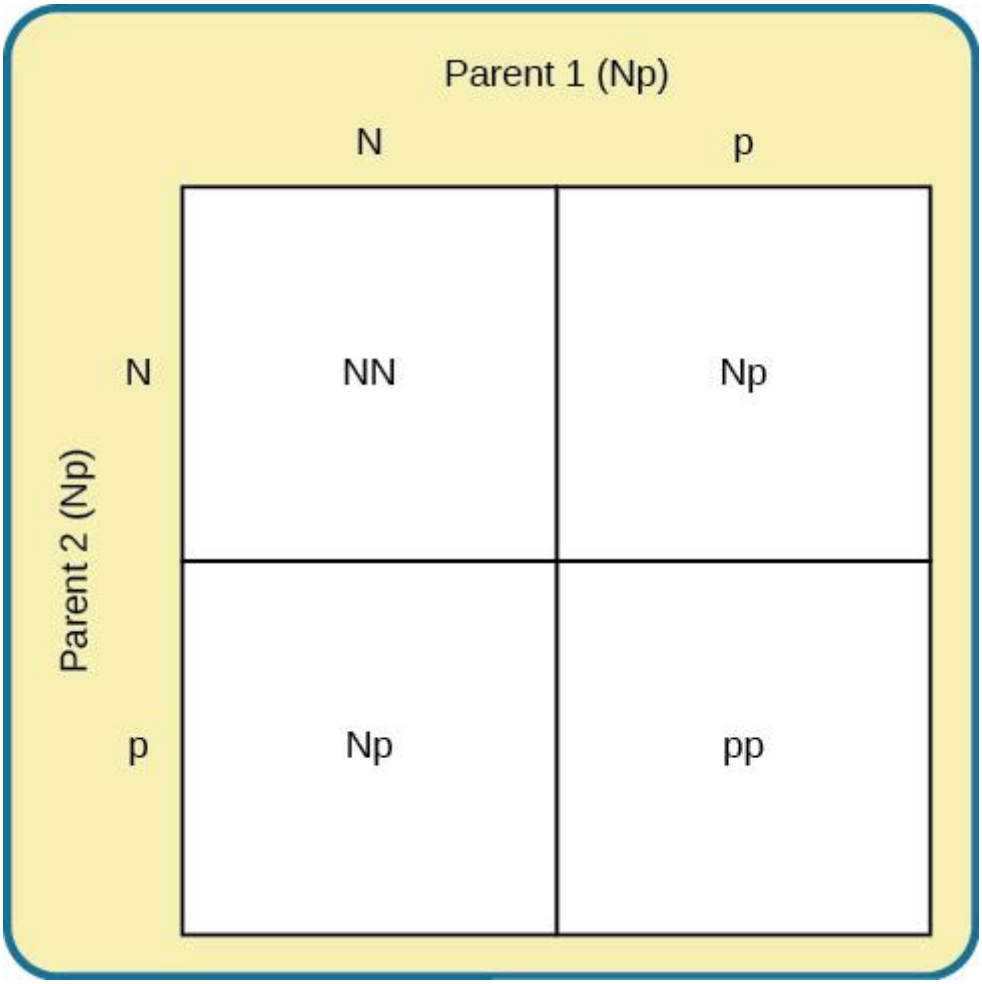

Figure 3.6 In this Punnett square, N represents the normal allele, and p represents the recessive allele that is associated with PKU. If two individuals mate who are both heterozygous for the allele associated with PKU, their offspring have a 25% chance of expressing the PKU phenotype.

Where do harmful genes that contribute to diseases like PKU come from? Gene mutations provide one source of harmful genes. A **mutation** is a sudden, permanent change in a gene. While many mutations can be harmful or lethal, once in a while, a mutation benefits an individual by giving that person an advantage over those who do not have the mutation. Recall that the theory of evolution asserts that individuals best adapted to their particular environments are more likely to reproduce and pass on their genes to future generations. In order for this process to occur, there must be competition—more technically, there must be variability in genes (and resultant traits) that allow for variation in adaptability to the environment. If a population consisted of identical individuals, then any dramatic changes in the environment would affect everyone in the same way, and there would be no variation in selection. In contrast, diversity in genes and associated traits allows some individuals to perform slightly better than others when faced with environmental change. This creates a distinct advantage for individuals best suited for their environments in terms of successful reproduction and genetic transmission.

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Human Diversity

This chapter focuses on biology. Later in this course you will learn about social psychology and issues of race, prejudice, and discrimination. When we focus strictly on biology, race becomes a weak construct. After the sequencing of the human genome at the turn of the millennium, many scientists began to argue that race was not a useful variable in genetic research and that its continued use represents a potential source of confusion and harm. The racial categories that some believed to be helpful in studying genetic diversity in humans are largely irrelevant. A person's skin tone, eye color, and hair texture are functions of their genetic makeups, but there is actually more genetic variation within a given racial category than there is between racial categories. In some cases, focus on race has led to difficulties with misdiagnoses and/or under-diagnoses of diseases ranging from sickle cell anemia to cystic fibrosis. Some argue that we need to distinguish between ancestry and race and then focus on ancestry. This approach would facilitate greater understanding of human genetic diversity (Yudell, Roberts, DeSalle, & Tishkoff, 2016).

Gene-Environment Interactions

Genes do not exist in a vacuum. Although we are all biological organisms, we also exist in an environment that is incredibly important in determining not only when and how our genes express themselves, but also in what combination. Each of us represents a unique interaction between our genetic makeup and our environment; range of reaction is one way to describe this interaction. **Range of reaction** asserts that our genes set the boundaries within which we can operate, and our environment interacts with the genes to determine where in that range we will fall. For example, if an individual's genetic makeup predisposes them to high levels of intellectual potential and they are reared in a rich, stimulating environment, then they will be more likely to achieve full potential than if they were raised under conditions of significant deprivation. According to the concept of range of reaction, genes set definite limits on potential, and environment determines how much of that potential is achieved. Some disagree with this theory and argue that genes do not set a limit on a person's potential with reaction norms being determined by the environment. For example, when individuals experience neglect or abuse early in life, they are more likely to exhibit adverse psychological and/or physical conditions that can last throughout their lives. These conditions may develop as a function of the negative environmental experiences in individuals from dissimilar genetic backgrounds (Miguel, Pereira, Silveira, & Meaney, 2019; Short & Baram, 2019).

Another perspective on the interaction between genes and the environment is the concept of **genetic environmental correlation**. Stated simply, our genes influence our environment, and our environment influences the expression of our genes (Figure 3.7). Not only do our genes and environment interact, as in range of reaction, but they also influence one another bidirectionally. For example, the child of an NBA player would probably be exposed to basketball from an early age. Such exposure might allow the child to realize their full genetic, athletic potential. Thus, the parents' genes, which the child shares, influence the child's environment, and that environment, in turn, is well suited to support the child's genetic potential.

Figure 3.7 Nature and nurture work together like complex pieces of a human puzzle. The interaction of our environment and genes makes us the individuals we are. (credit "puzzle": modification of work by Cory Zanker)

In another approach to gene-environment interactions, the field of **epigenetics** looks beyond the genotype itself and studies how the same genotype can be expressed in different ways. In other words, researchers study how the same genotype can lead to very different phenotypes. As mentioned earlier, gene expression is often influenced by environmental context in ways that are not entirely obvious. For instance, identical twins share the same genetic information (**identical twins** develop from a single fertilized egg that split, so the genetic material is exactly the same in each; in contrast, **fraternal twins** usually result from two different eggs fertilized by different sperm, so the genetic material varies as with non-twin siblings). But even with identical genes, there remains an incredible amount of variability in how gene expression can unfold over the course of each twin's life. Sometimes, one twin will develop a disease and the other will not. In one example, Aliya, an identical twin, died from cancer at age 7, but her twin, now 19 years old, has never had cancer. Although these individuals share an identical genotype, their phenotypes differ as a result of how that genetic information is expressed over time and through their unique environmental interactions. The epigenetic perspective is very different from range of reaction, because here the genotype is not fixed and limited.

LINK TO LEARNING

Watch this video about the epigenetics of twin studies to learn more.

Genes affect more than our physical characteristics. Indeed, scientists have found genetic linkages to a number of behavioral characteristics, ranging from basic personality traits to sexual orientation to spirituality (for examples, see Mustanski et al., 2005; Comings, Gonzales, Saucier, Johnson, & MacMurray, 2000). Genes are also associated with temperament and a number of psychological disorders, such as depression and schizophrenia. So while it is true that genes provide the biological blueprints for our cells, tissues, organs, and body, they also have a significant impact on our experiences and our behaviors.

Let’s look at the following findings regarding schizophrenia in light of our three views of gene-environment interactions. Which view do you think best explains this evidence?

In a 2004 study by Tienari and colleagues, adoptees whose biological mothers had schizophrenia *and* who had been raised in a disturbed family environment were much more likely to develop schizophrenia or another psychotic disorder than were any of the other groups in the study:

- Of adoptees whose biological mothers had schizophrenia (high genetic risk) and who were raised in disturbed family environments, 36.8% were likely to develop schizophrenia.
- Of adoptees whose biological mothers had schizophrenia (high genetic risk) and who were raised in healthy family environments, 5.8% were likely to develop schizophrenia.
- Of adoptees with a low genetic risk (whose mothers did not have schizophrenia) and who were raised in disturbed family environments, 5.3% were likely to develop schizophrenia.
- Of adoptees with a low genetic risk (whose mothers did not have schizophrenia) and who were raised in healthy family environments, 4.8% were likely to develop schizophrenia.

The study shows that adoptees with high genetic risk were most likely to develop schizophrenia if they were raised in disturbed home environments. This research lends credibility to the notion that both genetic vulnerability and environmental stress are necessary for schizophrenia to develop, and that genes alone do not tell the full tale.