

# 16-1 Genes and Variation

Standards: Bio 7c, Bio 7d

## Vocabulary

*gene pool, relative frequency, single-gene trait, polygenic trait*

As Darwin developed his theory of evolution, he worked under a serious handicap. He didn't know how heredity worked! Although Mendel's work on inheritance in peas was published during Darwin's lifetime, its importance wasn't recognized for decade. This lack of knowledge left two big gaps in Darwin's thinking .First, he had no idea how heritable traits pass from one generation to the next. Second, although variation in heritable traits was central to Darwin's theory, he had no idea how that variation appeared.

Evolutionary biologists connected Mendel's work to Darwin's during the 1930s. By then, biologists understood that genes control heritable traits. They soon realized that changes in genes produce heritable variation on which natural selection can operate. Genes became the focus of new hypotheses and experiments aimed at understanding evolutionary change. Another revolution in evolutionary thought began with Watson and Cricks' studies on DNA. Their model of the DNA molecule helped evolutionary biologists because it demonstrated the molecular nature of mutation and genetic variation.

Today, molecular techniques are used to test hypotheses about how heritable variation appears and how natural selection operates on that variation. AS you will learn in this chapter, fitness, adaption, species, and evolutionary change are now defined in genetic terms. WE understand how evolution works better than Darwin ever could, beginning with heritable variation.

## How Common Is Genetic Variation?

We now know that many genes have at least two forms, or alleles. Animals such as horses, dogs, and mice often have several alleles for traits such as body size or coat color. Plants, such as peas, often have several alleles for flower color. All organisms have additional genetic variation that is "invisible" because it involves small differences in biochemical processes. In addition, an individual organism is heterozygous for many genes. An insect may be heterozygous for as many as 15 percent of its genes. Individual fishes, reptiles, and mammals are typically heterozygous for between 4 and 8 percent of their genes.

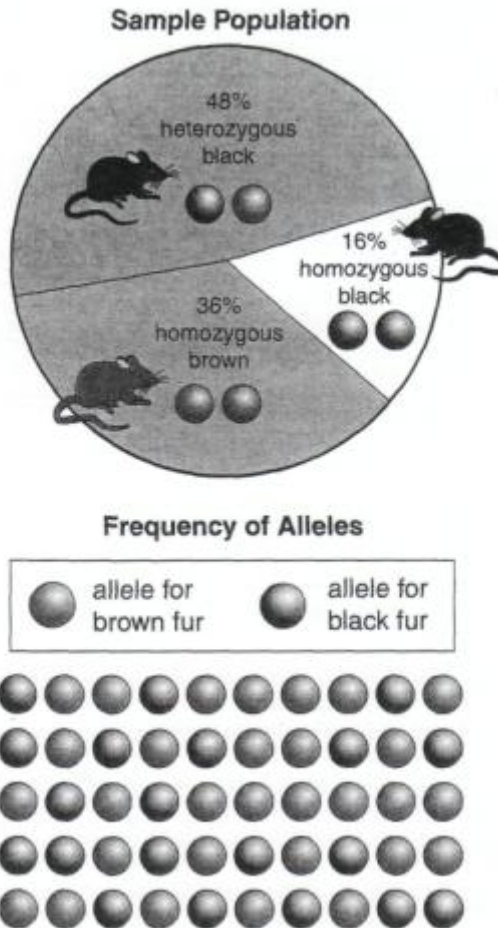
## Variation and Gene Pools

Genetic variation is studied in population. A population is a group of individuals of the same species that interbreed. Because members of a population interbreed, they share a common group of genes called a gene pool. A **gene pool** consists of all genes, including all the different alleles that are present in a population.

The **relative frequency** of an allele is the number of times that the allele occurs in a gene pool, compared with the number of times other alleles for the same gene occur. Relative frequency is often expressed as a percentage. For example, in the mouse population in Figure 16-2, the relative frequency of the dominant B allele (black fur) is 40 percent, and the relative frequency of the recessive B allele (brown fur) is 60 percent. The relative

frequency of an allele has nothing to do with whether the allele is dominant or recessive. In this particular mouse population, the recessive allele occurs more frequently than the dominant allele.

Gene pools are important to evolutionary theory, because evolution involves changes in populations over time. **In genetic terms, evolution is any change in the relative frequency of alleles in a population.** For example, if the relative frequency of the B allele in the mouse population changed over time to 30 percent, the population is evolving.



✓Checkpoint: What is a gene pool?

## Sources of Genetic Variation

Biologists can now explain how variation is produced. **The two main sources of genetic variation are mutations and the genetic shuffling that results from sexual reproduction.**

**Mutations** A mutation is any change in a sequence of DNA. Mutations can occur because of mistakes in the replication of DNA or as a result of radiation or chemicals in the environment. Mutations do not always affect an organism's phenotype. For example, a DNA codon altered from GGA to GGU will still code for the same amino acid, glycine. That mutation has no effect on phenotype. Many mutations do produce changes in phenotype,

however. Some can affect an organism's fitness, or its ability to survive and reproduce in its environment. Other mutations may have no effect on fitness.

**Gene Shuffling** Mutations are not the only source of heritable variation. You do not look exactly like your biological parents, even though they provided you with all your genes. You probably look even less like any brothers or sisters you may have. Yet, no matter how you feel about your relatives, mutant genes are not primarily what makes them so different from you. Most heritable differences are due to gene shuffling that occurs during the production of gametes. Recall that each chromosome of a homologous pair moves independently during meiosis. As a result, the 23 pairs of chromosomes found in humans can produce 8.4 million different combinations of genes!

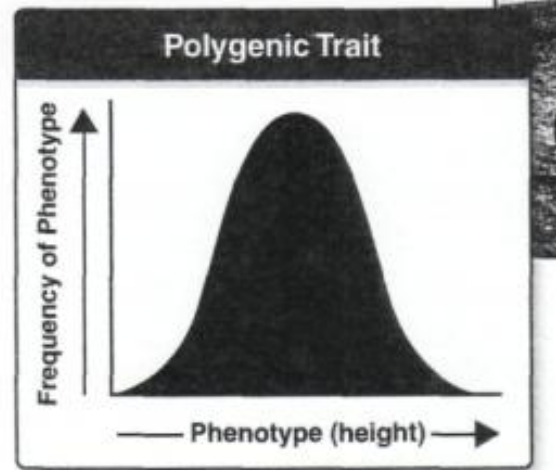
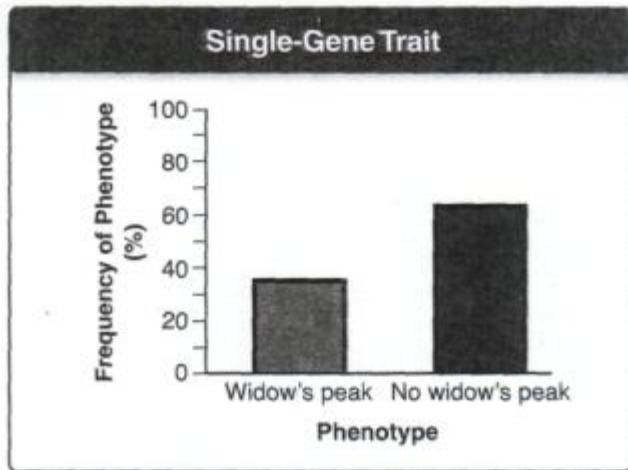
Another process, crossing-over, also occurs during meiosis. Crossing-over further increases the number of different genotypes that can appear in offspring. Recall that a genotype is an organism's genetic makeup. When alleles are recombined during sexual reproduction, they can produce dramatically different phenotypes. Thus, sexual reproduction is a major source of variation within many populations.

Sexual reproduction can produce many different phenotypes, but it does not change the relative frequency of alleles in a population. To understand why, compare a population's gene pool to a deck of playing cards. Each card represents an allele found in the population. The exchange of genes during gene shuffling is similar to shuffling a deck of cards. Shuffling leads to different types of hands, but it can never change the relative number of aces, kings, or queens in the deck. The probability of drawing an ace off the top of the deck will always be 4 in 52, or one thirteenth ( $4/52 = 1/13$ ). No matter how many times you shuffle the deck, this probability will remain the same. Similarly, sexual reproduction produces many different combinations of genes, but in itself it does not alter the relative frequencies of each type of allele in a population.

√*Checkpoint: What are the sources of heritable variation?*

## Single-Gene and Polygenic Traits

Heritable variation can be expressed in a variety of ways. **The number of phenotypes produced for a given trait depends on how many genes control the trait.** Among humans, a widow's peak — a downward dip in the center of the hairline — is a **single-gene trait**. It is controlled by a single gene that has two alleles. The allele for a widow's peak is dominant over the allele for a hairline with no peak. As a result, variation in this gene leads to only two distinct phenotypes, as shown.



As you can see, the frequency of phenotypes caused by this single gene is represented on the bar graph. This graph shows that the presence of a widow's peak may be less common in a population than the absence of a widow's peak, even though the allele for a widow's peak is the dominant form. In real populations, phenotypic ratios are determined by the frequency of alleles in the population as well as by whether the alleles are in the dominant or recessive form. Allele frequencies may not match Mendelian ratios.

Many traits are controlled by two or more genes and are, therefore, called polygenic traits. Each gene of a polygenic trait often has two or more alleles. As a result, one polygenic trait can have many possible genotypes and phenotypes.

Height in humans is one example of a polygenic trait. You can sample phenotypic variation in this trait by measuring the height of all the students in your class. You can then calculate the average height of this group.

Many students will be just a little taller or shorter than average. Some of your classmates, however, will be very tall or very short. If you graph the number of individuals of each height, you may get a graph similar to the one in Figure 16-4. The symmetrical bell-like shape of this curve is typical of polygenic traits. A bell-shaped curve is also called a normal distribution.

## 16-1 Section Assessment

1. In genetic terms, what indicates that evolution is occurring in a population?
2. What two processes can lead to inherited variation in populations?
3. How does the range of phenotypes differ between single-gene traits and polygenic traits?
4. What is a gene pool? How are allele frequencies related to gene pools?
5. Evaluate the significance of mutations to the process of biological evolution. (Hint: How does mutation affect genetic variation?)