11.1 Population Genetics

EXPECTATIONS

- **Analyze evolutionary mechanisms and their effects on biodiversity and extinction.**
- **Understand how alleles are distributed in a population.**
- **Understand how allele ratios reflect selective pressure in a population.**

Evolution can be divided into macro-evolution and micro-evolution. **Macro-evolution** is evolution on a grand scale; it is large evolutionary change such as the evolution of new species from a common ancestor or the evolution of one species into two. (You will learn more about the evolution of new species in Chapter 12.) The modern camel, for example, evolved over 65 million years from a small ancestor that was not much larger than a rabbit. This long, visible sequence of changes and the categorization of organisms (extinct and living) in relation to one another are examples of macro-evolution. Figure 11.1 shows the sequence that paleontologists propose for the evolutionary path of the modern camel. (Ancestral camels actually evolved in North America, and then expanded their range to include parts of Asia and Africa.)

Micro-evolution is the change in the gene frequencies within a population over time. It is evolution *within* a species, or evolution on a small scale. For example, adaptation by natural selection is an example of evolutionary change within a species, or micro-evolution. As these changes accumulate, they can lead to the formation of a new species.

This chapter focusses on micro-evolution and the mechanisms that result in genetic variation within a population.

Heredity and Evolution

While Darwin's *The Origin of Species* convinced most biologists that species could change over time, Darwin's mechanism for change — natural selection — took longer to gain acceptance. Part of the difficulty in explaining the mechanism of natural selection was that there needed to be a plausible explanation for how traits could be inherited. If variation within a species was necessary for natural selection, what was the ultimate source of this variation?

In Darwin's theory of natural selection, new variants of species arise continually in populations. Some variants thrive and produce more offspring,

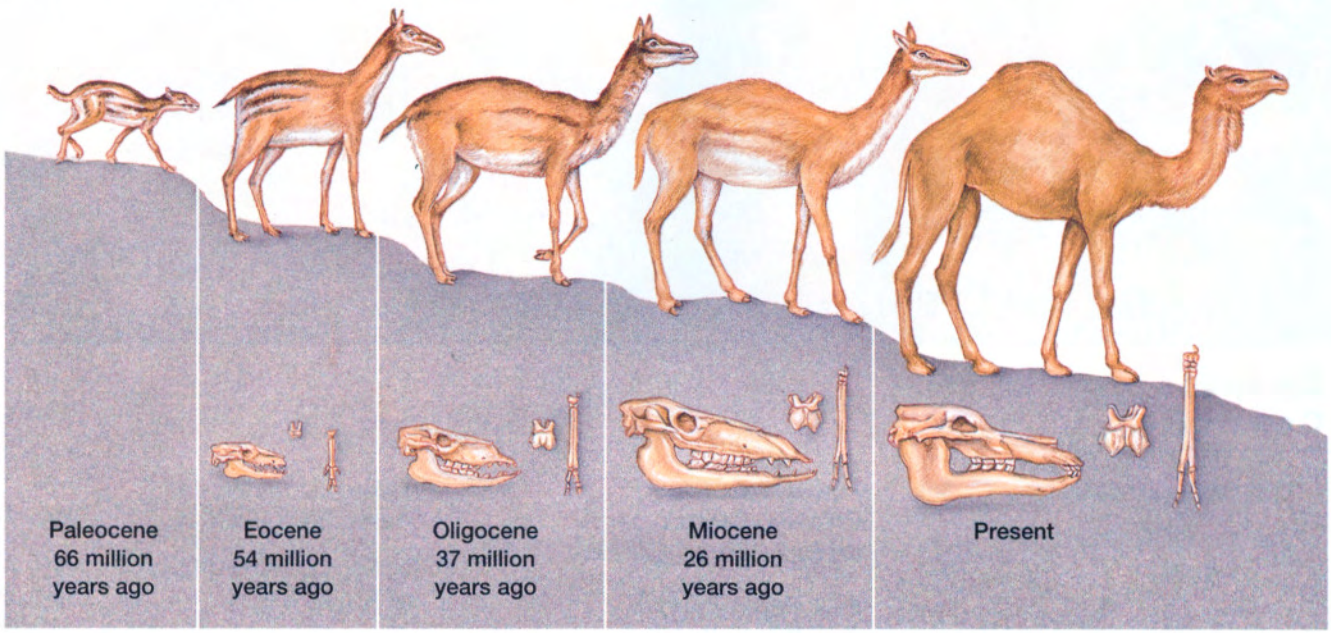

Figure 11.1 Paleontologists have used fossils to trace the evolution of the modern camel.

thus slowly leading to change in a population (this may even lead to new species over time), while other variants die off because they cannot thrive in the environment. This idea did not mesh with the ideas about inheritance at that time, which said that characteristics were blended and an offspring was an "average" of its parents. (For example, according to blended inheritance, the offspring of a plant with a red flower and a plant with a white flower would be pink. This offspring would then pass on the pink colour to its offspring. In reality, this is not always the case.) What was needed to support natural selection was an understanding of how chance variations arise in a population and how these variations are passed from parents to offspring.

Part of this missing information needed to explain inheritance and support the idea of evolution by natural selection was actually discovered during Darwin's lifetime. Gregor Mendel, an Austrian monk who is shown in Figure 11.2, conducted experiments with pea plants in the 1850s, and his work provided the basis for an explanation of inheritance. His experiments showed that, in contrast to the idea of blended inheritance, parents pass on discrete factors of inheritance, which he called genes. Mendel showed that genes do not blend in the offspring; genes retain their characteristics when they are passed to the offspring. Mendel's work and the subsequent work of others on inheritance would eventually help support the idea of natural selection by showing how the variation created through the mechanisms of heredity is the raw material on which natural selection acts.

Figure 11.2 Gregor Mendel (1822–1884) conducted experiments that explained the inheritance of characteristics.

Mendel's work was some of the first that helped to explain mechanisms of inheritance. But his work was not read by Darwin, and it would be several decades before ideas about inheritance were used to help explain natural selection. In the late nineteenth and early twentieth centuries, there was a growing interest in genetics. In the 1930s, a new field of science emerged — population genetics. As scientists began to broaden their understanding of genetics, they demonstrated that there is substantial genetic variation within populations. They showed that variations could arise in populations through changes, or **mutations**, in genes. A mutation is a permanent change in the genetic material of an organism. (Refer back to Chapter 9, section 9.1 to review mutations.) It was recognized that mutations provide the genetic variation within a population. Evolution, therefore, depends on both random genetic mutation (which provides variation) and mechanisms such as natural selection. (You will learn more about mutations in section 11.3.)

Scientists, including geneticist Theodosius Dobzhansky, biogeographer and taxonomist Ernst Mayr, paleontologist George Gaylord Simpson, and botanist G. Ledyard Stebbins, combined ideas from their fields of study with Darwin's ideas about natural selection and the current understanding of inheritance to develop a revised theory of evolution. This modification to evolutionary theory, and the meshing of Mendel's and Darwin's ideas, was called the **modern synthesis**.

Reviewing the Language of Genetics

To understand and discuss genetic variation, it is important to review certain terms. **Alleles** are alternate forms of a gene. In humans, for example, there are three alleles — I^A, I^B, and i — that determine whether an individual has A, B, AB, or O blood type. Since individuals generally have two sets of chromosomes — one received from the male parent and one received from the female parent — there are two alleles for every gene at every locus. (A **locus** [plural loci] is the location of a gene on a chromosome.) So, humans could be I^AI^A, I^AI^B, I^Ai, I^BI^B, I^Bi, or ii at the locus for blood group. If the

two alleles at a locus are identical (for example, I^AI^A or *ii*), the individual is called **homozygous** for that characteristic. An individual with two different alleles at the locus (for example, I^AI^B) is called **heterozygous**. The three blood type alleles, I^A, I^B, or *i*, exist in the population, but no single person can have all three. In some populations, the allele possibilities are even greater, and far exceed the two possible alleles any human can have.

If the two alleles inherited from parents are different, one of them (the **dominant allele**) will be fully expressed in the organism's appearance and therefore will become the phenotype. (Note that dominant in this sense does not mean that this allele is somehow better. Trait for the dominant allele is simply the one that is always expressed in an individual.) The other allele, the **recessive allele**, has no noticeable effect on the organism's appearance, but it remains as part of the genotype of the organism. Figure 11.3 shows a cross between a pure purple-flowered pea plant and a pure white-flowered pea plant. The alleles for colour are W and w. Since W is the dominant allele, the flowers can only be white when the two alleles are both recessive (that is, ww).

The **genotype**, or genetic make-up, of an *individual* remains constant throughout its life. However, over time, the alleles within a *population* may change. New alleles may arise and may be recombined, thus producing individuals with novel **phenotypes**. Phenotypes are the physical and physiological traits of an organism. (Physical in this sense refers to how the organism with this trait appears.) A phenotype of an individual can be the product of both the environment and heredity. For example, environmental factors such as disease, crowding, injury, or the availability of food can all affect the appearance of an individual. But these acquired characteristics are not heritable; that is, they are not passed on to the next generation. Because of dominant and recessive alleles, an organism's appearance does not always reflect its genetic make-up. For example, Figure 11.4 shows a cross between two pea plants that have the alleles W or w at the locus for colour. The genotypes WW and Ww both result in a purple flower, while the genotype ww results in a white flower. Table 11.1 summarizes how genotype is related to phenotype.

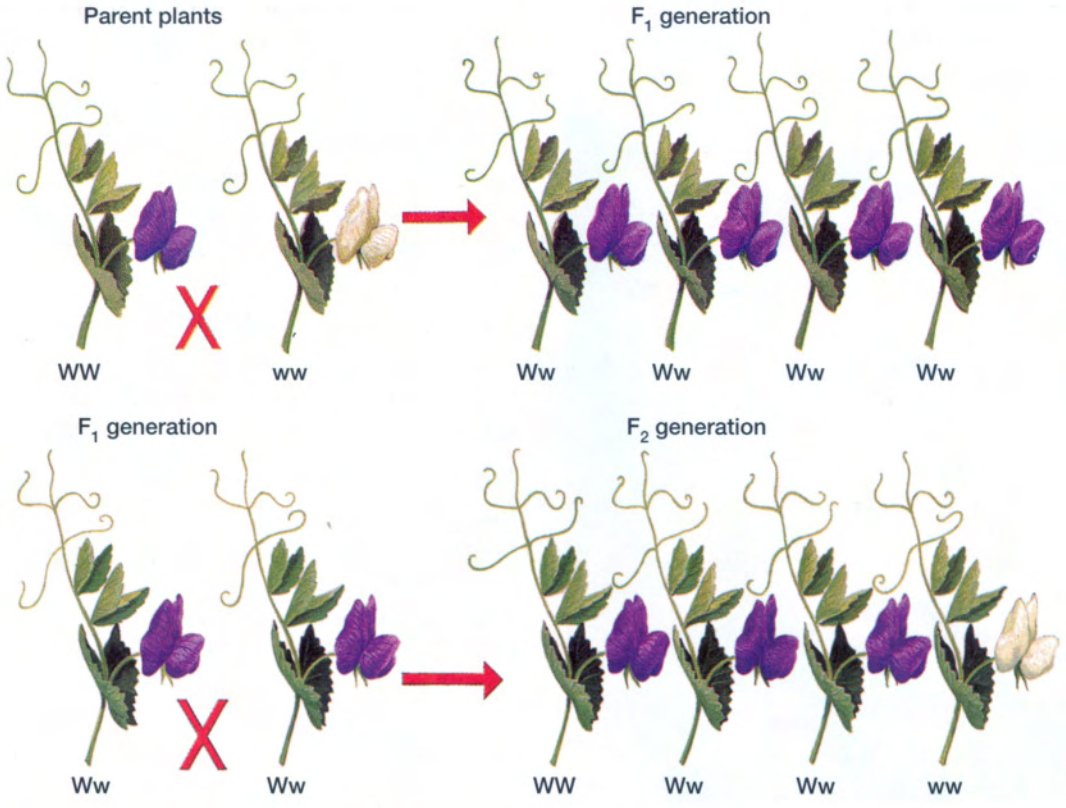

Figure 11.3 Two generations resulting from the cross of a purple-flowered pea plant and a white-flowered pea plant.

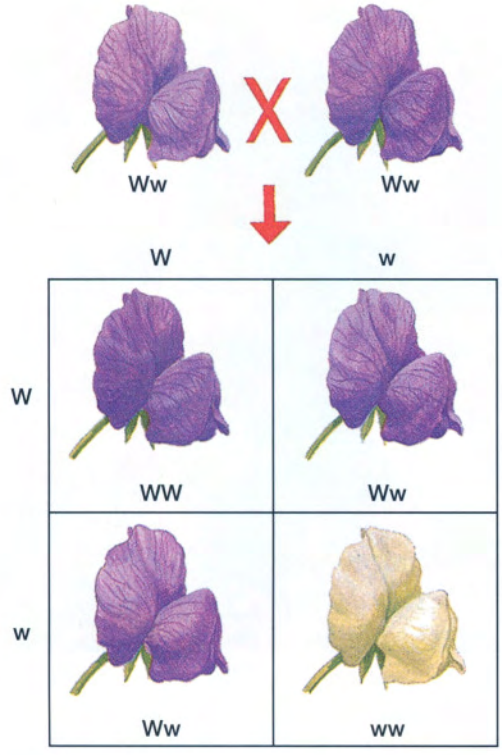

Figure 11.4 The result of a cross of two pea plants is shown in this Punnett square.

Not all traits are purely dominant or purely recessive. Sometimes neither of the alleles controlling a trait are dominant. In this case, blending of the two traits can occur. This is called **incomplete dominance**. Figure 11.5 gives an example of how incomplete dominance can occur. White or red flowers are homozygous, while pink flowers are heterozygous.

Occasionally both alleles for a trait may be dominant. These alleles are said to be **co-dominant** and both alleles are expressed in the heterozygous individual. In some varieties of chickens, for example, two alleles for a trait may be expressed equally. A black rooster crossed with a white hen produces offspring that have some black feathers and some white feathers.

Table 11.1
Genotype and phenotype in peas with alleles W and w

Genotype	Genotype	Phenotype
WW	homozygous dominant	purple flowers
Ww	heterozygous	purple flowers
ww	homozygous recessive	white flowers

Population Genetics

A **population** is a localized group of a single species occupying a particular area. For example, the field of lilies in Figure 11.6A on page 368 is a different population from a field of lilies in an adjacent valley. The two populations are not completely isolated (since pollinating insects may travel between them), but it is more likely that members of the same population will interbreed to produce the next generation. The same is true for the pond of frogs in Figure 11.6B. Although it is not out of the question that these frogs would mate with frogs from a nearby pond, it is more likely they would mate with individuals that live in the same pond.

Monarch butterflies (such as those shown on page 363) are all from the same species, yet there are distinct populations within this species. When the butterflies migrate in winter, they travel in huge flocks and become mixed. In their summer breeding grounds, however, they have strong family groupings. Genetic mixing during migration and in their winter habitat ensures that the species does not begin to diverge into two or more species. However,

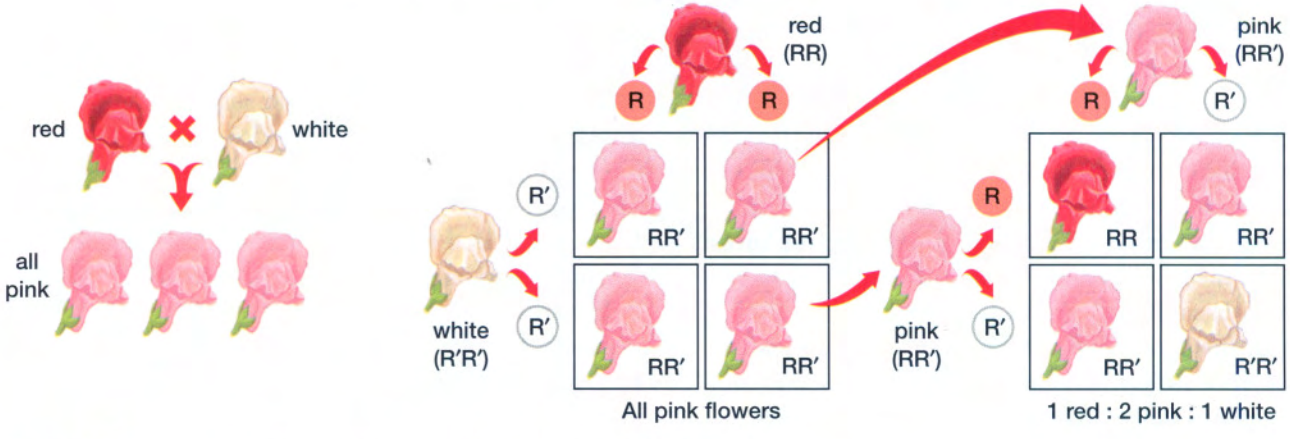

Figure 11.5 Flower colour in the snapdragon is an example of incomplete dominance. Pink flowers are heterozygous (*RR′*), where neither allele is dominant.

Figure 11.6 A field of glacier lilies and a pond of frogs are each considered populations.

the genetic similarity of the smaller summer groupings creates diversity among local populations.

All of the genes in a population or, more specifically, all of the alleles at all gene loci in all individuals of the population, make up the population's gene pool. There can be genetic variation both within individuals (when they are heterozygous for alleles) and within populations. For example, most plants within a population have more than one allele, or are **polymorphic**, at 45 percent of the loci. As well, individual plants are likely to be heterozygous at about 15 percent of their loci. A polymorphic population (with organisms exhibiting different phenotypes and genotypes) and heterozygous individuals contribute to the level of genetic variability within a population. When all members of a population are homozygous for the same allele, that allele is said to be **fixed** in the gene pool. In most circumstances, however, there are two or more alleles for a gene and each exists with a relative frequency within the population. The unique combination of alleles in individuals provides the variation within a population.

Scientists can use the technique of **electrophoresis** to help measure genetic variation within populations. Recall from Chapter 9 (section 9.2) that in the process of electrophoresis, samples of DNA from individuals are placed in a special gel that is then placed in a solution and connected to an electrical circuit. The DNA fragments move through the gel at varying speeds and the resulting pattern of bands — called the DNA fingerprint — is stained and analyzed. Biologists can use this technique to look at the variability of genes (and, consequently, genetic variation) in the population. To do this they compare the samples from different individuals within a population to calculate the percentage of loci that are polymorphic. The more sites that are polymorphic, the greater the genetic variety within the population.

Polymerase chain reactions (PCR) are also used by evolutionary biologists. (Recall that PCR was introduced on page 287.) PCR techniques are used to amplify (generate multiple copies of) DNA from small samples. For example, even minute samples of DNA gathered from mummified organisms or fossils can be copied using PCR techniques. Then, the DNA can be analyzed and compared with DNA sequences of other organisms to help determine evolutionary relationships. Electrophoresis and PCR techniques can be used to sequence and analyze DNA taken from long-dead, or even long-extinct, organisms. For example, DNA has been taken from a 76 000-year-old mummified human brain, fossilized bacteria, and a 40 000-year-old frozen woolly mammoth. This information will help determine the evolutionary history of organisms, because the relatedness of species can be reflected in DNA and proteins. Species that are closely related share a greater proportion of their DNA sequences and proteins.

BIO FACT

In the blood hemoglobin molecule of 146 amino acids, humans and gorillas differ by just one amino acid. Humans and frogs, however, differ by 67 amino acids.

Population geneticists study the frequencies of alleles and genotypes in populations. The study of population genetics is important to the study of micro-evolution because changes in the genetic variability within the population can be used to determine if a population is undergoing micro-evolution. To illustrate how frequencies of alleles and genotypes can be calculated, let's consider a hypothetical population of 400 field mice that are either white or black (see Figure 11.7). The allele for black, A, is dominant to the allele for white, a. (For this example we will assume there are only two alleles for this locus.) In this population of mice, 364 are black and 36 are white. Of the black mice, 196 are homozygous dominant (AA) and 168 are heterozygous (Aa). The 36 white mice are homozygous recessive, aa. Since these mice inherit one set of chromosomes from each parent (that is, they are **diploid**), there are a total of 800 copies of genes for fur colour in the population of 400 field mice. The dominant allele (A) accounts for 560 of these genes ($196 \times 2 = 392$ for AA mice and $168 \times 1 = 168$ for Aa mice). The recessive allele (a) accounts for 240 of these genes ($36 \times 2 = 72$ for aa mice and $168 \times 1 = 168$ for Aa mice). The overall frequency of the A allele in the gene pool of this population is $560/800 = 0.7 = 70\%$, and the frequency of the a allele is 0.3 or 30%. (**Frequency** is the number of occurrences of a particular allele in a population divided by the total number of alleles in the population.) The genotypic frequencies in this population are: AA = 0.49 (196 out of 400 mice), Aa = 0.42 (168/400), and aa = 0.09 (36/400). The frequencies of alleles and genotypes are called the population's **genetic structure**.

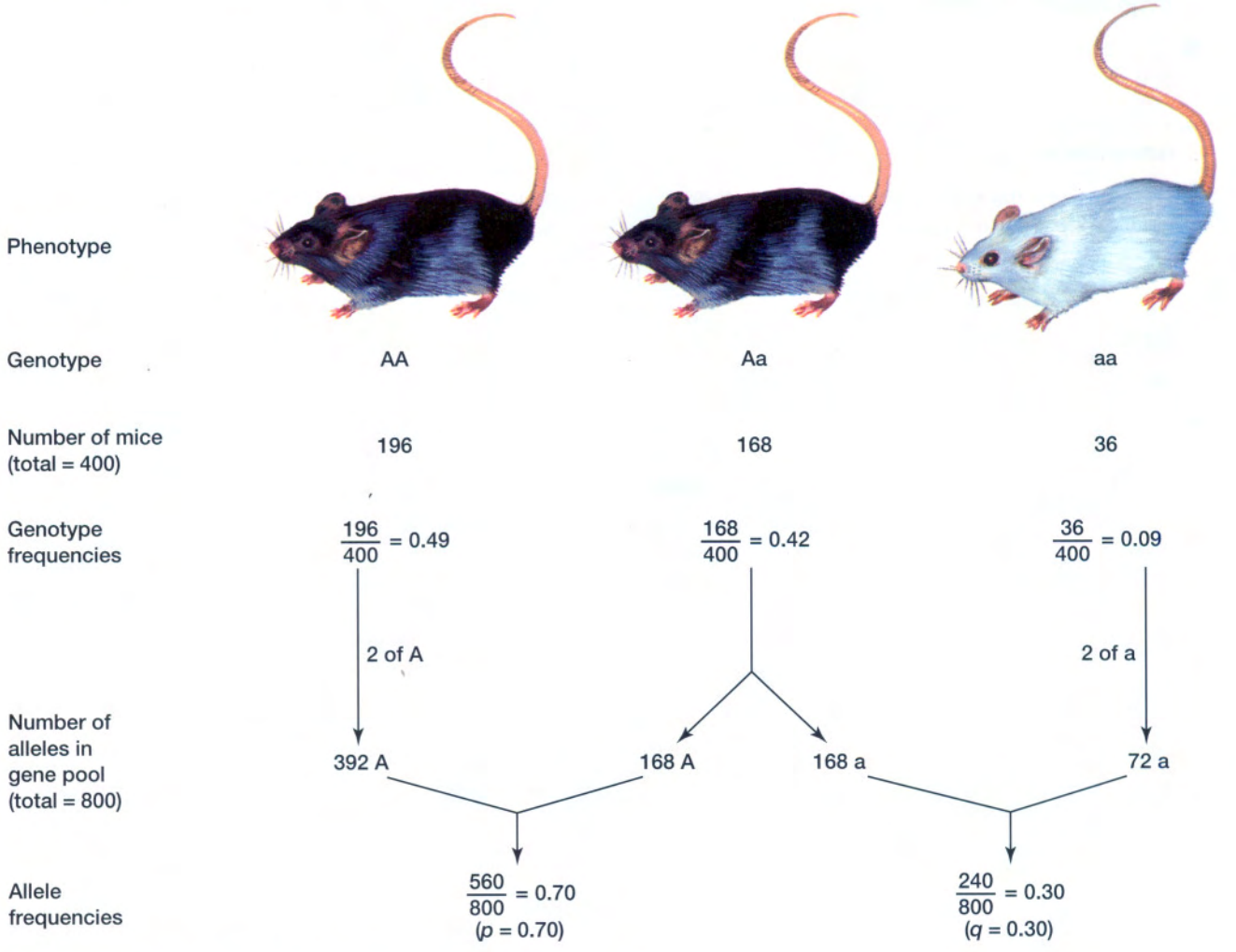

Figure 11.7 Genetic structure of a parent population of field mice

Practice Problems

1. Calculate the allele frequency and genotype frequency in a population of 500 flowers in which 275 are homozygous dominant, 137 are heterozygous, and 88 are homozygous recessive.
2. In a population of 500 plants with purple flowers and white flowers, the genotype frequency is AA = 0.64, Aa = 0.32, and aa = 0.04. Calculate the frequency of each allele in the population. If A is the allele for purple and a is the allele for white, calculate the numbers of each colour of plant. Copy the following chart into your notebook and fill in the data.

phenotype			
genotype			
number			
genotype frequencies			
number of alleles in gene pool			
allele frequencies			

SECTION REVIEW

1. K/U Distinguish between macro-evolution and micro-evolution. Give an example of each.
2. K/U Explain how genetic variation and micro-evolution are related.
3. K/U Explain how variations within a species are affected by natural selection.
4. K/U Use the word "population" to explain local differences in a species.
5. K/U Describe the similarities and differences between the following pairs:
   (a) allele and gene
   (b) phenotype and genotype
   (c) dominant and recessive
   (d) homozygous and heterozygous
6. I Assume that a white animal is crossed with three other animals of the same species, A, B, and C. (For this example we will assume there are only two alleles for this locus.) Animal A is brown and produces offspring A′, which is also brown. Animal B is white and produces offspring B′, which is brown. Animal C is brown and produces offspring C′, which is white. Give the genotypes and phenotypes of all seven animals. Show how you came to this conclusion.
7. K/U Explain how polymorphic populations and heterozygous individuals contribute to the level of genetic variety in a population.
8. C Describe the genetic structure (genotype and allele frequency) using a chart or table for a population of 300 frogs. The frogs are either spotted or spot-less. The allele for spots, A, is dominant to the allele for no spots, a. In this population, 240 frogs have spots and 60 are spot-less. Of the 240 frogs with spots, 200 are homozygous and 40 are heterozygous.
9. I If a group of scientists is trying to determine how a particular fossil of a woolly mammoth is related to an animal on Earth today, what techniques could they use? How could they determine relatedness?