3

Basic Principles of Heredity

Blond hair occurs in 5%–10% of dark-skinned Solomon Islanders. Blond hair in this group is a recessive trait and has a different genetic basis from blond hair in Europeans. [© Anthony Asael/Danita Delimont/Alamy.]

The Genetics of Blond Hair in the South Pacific

A thousand miles northeast of Australia lies an ancient chain of volcanic and coral islands known as the Solomons (**Figure 3.1**). The Solomon Islands were first inhabited some 30,000 years ago, when Neanderthals still roamed northern Europe. Today, the people of the Solomons are culturally diverse but consist largely of Melanesians, a group that also inhabits other South Pacific islands. Most people from the Solomon Islands have dark skin. Remarkably, 5%–10% also have strikingly blond hair; in fact, people of the Solomon Islands have the highest frequency of blond hair outside Europe.

How did the Solomon Islanders get their blond hair? A number of hypotheses have been proposed over the years. Some suggested that the blond islanders had naturally dark hair that was bleached by the sun and salt water. Others proposed that the blond hair color was caused by diet. Still others suggested that it was the result of genes for blond hair left by early European explorers.

The mystery of the blond Solomon Islanders was solved in 2012 by geneticists Eimear Kenny and Sean Myles and their colleagues. Their research demonstrated that blond hair on the islands is, in fact, caused by a gene, but not one left by Europeans—blond hair in Solomon Islanders and in Europeans have completely separate evolutionary origins.

To search for the origin of blond hair among the people of the Solomon Islands, the geneticists collected saliva and hair samples from over 1200 people on the islands, from which they extracted DNA. In a type of analysis known as a genome-wide association study, they looked for statistical associations between the presence of blond hair and thousands of genetic variants scattered across the genome. Right away, they detected a strong correlation between the presence of blond hair and a particular genetic variant located on the short arm of chromosome 9. This region of chromosome 9

3.1 Map of the Solomon Islands.

contains the tyrosinase-related protein 1 (*TYRP1*) gene, which encodes an enzyme known to play a role in the production of melanin and to affect pigmentation in mice. The researchers found a single base difference between the DNA of islanders with blond hair and that of islanders with dark hair: the blonds had a thymine (T) base instead of a cytosine (C) base in their *TYRP1* gene.

Further research showed that blond hair among Solomon Islanders is a recessive trait, meaning that blonds carry two copies of the blond version of the gene (*TT*)—one inherited from each parent. Dark hair is dominant: dark-haired islanders carry either one (*CT*) or two (*CC*) copies of the dark-hair version of the gene. Thus, many dark-haired islanders are heterozygous, carrying a hidden copy of the blond gene that can be passed on to their offspring. A DNA analysis of 900 Solomon Islanders demonstrated that over 40% of dark-haired islanders carry a blond gene. Interestingly, the C-to-T mutation in the *TYRP1* gene that causes blond hair in Solomon Islanders is rare outside the South Pacific, suggesting that the mutation arose independently within the Melanesian population. There is no evidence that the gene was inherited from Europeans.

THINK-PAIR-SHARE Question 1

The genetics of blond hair in Solomon Islanders differs from that in Europeans in other ways as well. In Europeans, variations in at least eight different genes have been associated with blond hair. In 2015, researchers examined one of these genes (called *KITLG*) and found that the mutation causing blond hair occurred not in the gene itself but in a region of DNA that affects the expression of the *KITLG* gene. The *KITLG* gene produces a protein that is involved in a number of functions, including melanocyte development and melanin synthesis.

THINK-PAIR-SHARE Questions 2 & 3

This chapter is about the principles of heredity: how genes—such as the one for blond hair among Solomon Islanders—are passed from generation to generation and how factors such as dominance influence their inheritance. The principles of heredity were first put forth by Gregor Mendel, so we begin this chapter by examining Mendel's scientific achievements. We then turn to simple genetic crosses in which a single characteristic is examined. We consider some techniques for predicting the outcome of genetic crosses, and then turn to crosses in which two or more characteristics are examined. We see how the principles applied to simple genetic crosses and the ratios of offspring they produce can serve as the key to understanding more complicated crosses. The chapter ends with a discussion of statistical tests for analyzing crosses.

Throughout this chapter, a number of concepts are interwoven: Mendel's principles of segregation and independent assortment, probability, and the behavior of chromosomes. These concepts might at first appear to be unrelated, but they are actually different views of the same phenomenon because the genes that undergo segregation and independent assortment are located on chromosomes. In this chapter, we examine these different views and clarify how they are related.

3.1 Gregor Mendel Discovered the Basic Principles of Heredity

It was in the early 1900s that the principles of heredity first became widely known among biologists. Surprisingly, these principles had been discovered some 44 years earlier by an Augustinian priest named Gregor Johann Mendel (1822–1884).

Mendel was born in what is now part of the Czech Republic. Although his parents were simple farmers with little money, he received a sound education and was admitted to the Augustinian monastery in Brno in September

1843. After graduating from seminary, Mendel became an ordained priest and was appointed to a teaching position in a local school. He excelled at teaching, and the abbot of the monastery recommended him for further study at the University of Vienna, which he attended from 1851 to 1853. There, Mendel enrolled in the newly opened Physics Institute and took courses in mathematics, chemistry, entomology, paleontology, botany, and plant physiology. It was probably there that Mendel acquired knowledge of the scientific method, which he later applied successfully to his genetic experiments.

After studying in Vienna, Mendel returned to Brno, where he taught school and began his experimental work with pea plants. He conducted breeding experiments from 1856 to 1863 and presented his results publicly at meetings of the Brno Natural Science Society in 1865. Mendel's paper based on these lectures was published in 1866. However, in spite of widespread interest in heredity, the effect of his research on the scientific community was minimal. At the time, no one seemed to have noticed that Mendel had discovered the basic principles of inheritance.

In 1868, Mendel was elected abbot of his monastery, and increasing administrative duties brought an end to his teaching and, eventually, to his genetic experiments. He died at the age of 61 on January 6, 1884, unrecognized for his contribution to genetics.

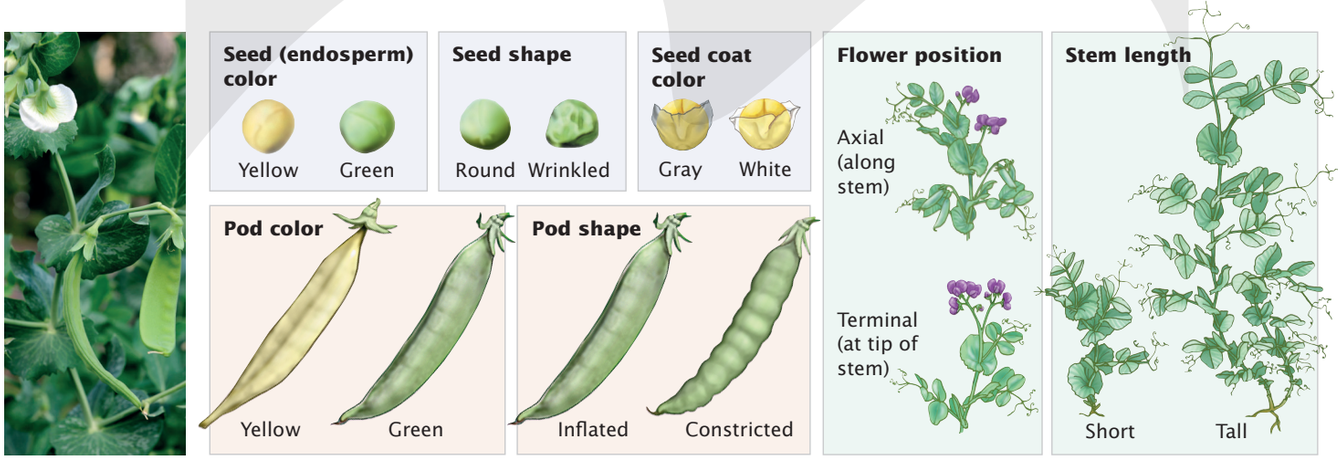
The significance of Mendel's discovery was not recognized until 1900, when three botanists—Hugo de Vries, Erich Tschermak von Seysenegg, and Carl Correns—began independently conducting similar experiments with plants and arrived at conclusions similar to those of Mendel. Coming across Mendel's paper, they interpreted their results in accord with his principles and drew attention to his pioneering work.

Mendel's Success

Mendel's approach to the study of heredity was effective for several reasons. Foremost was his choice of experimental subject, the pea plant *Pisum sativum* (**Figure 3.2**), which offered clear advantages for genetic investigation. The plant is easy to cultivate, and Mendel had the monastery garden and greenhouse at his disposal. Peas grow relatively rapidly, completing an entire generation in a single growing season. By today's standards, one generation per year seems frightfully slow—fruit flies complete a generation in 2 weeks and bacteria in 20 minutes—but Mendel was under no pressure to publish quickly and was able to follow the inheritance of individual characteristics for several generations. Had he chosen to work on an organism with a longer generation time—horses, for example—he might never have discovered the basis of inheritance. Pea plants also produce many offspring—their seeds—which allowed Mendel to detect meaningful mathematical ratios in the traits he observed in the progeny. The numerous varieties of peas available to Mendel were also crucial to his success because they differed in various traits and were genetically pure (homozygous). Mendel was therefore able to begin with plants of variable, known genetic makeup.

Much of Mendel's success can be attributed to the seven characteristics of pea plants that he chose for study (see Figure 3.2). He avoided characteristics that display a range of variation; instead, he focused on those that exist in two easily differentiated forms, such as white versus gray seed coats, round versus wrinkled seeds, and inflated versus constricted pods.

Finally, Mendel was successful because he adopted an experimental approach and interpreted his results by using mathematics. Unlike many earlier investigators who simply described the *results* of crosses, Mendel formulated *hypotheses* based on his initial observations and then



3.2 Mendel used the pea plant *Pisum sativum* in his studies of heredity. He examined seven characteristics that appeared in the seeds and in plants grown from the seeds. [© Charles Stirling/Alamy.]

conducted additional crosses to test his hypotheses. He kept careful records of the number of progeny possessing each trait and computed ratios of the different traits. He was adept at seeing patterns in detail and was patient and thorough, conducting his experiments for 10 years before attempting to write up his results. TRY PROBLEM 14

THINK-PAIR-SHARE Question 4

CONCEPTS

Gregor Mendel put forth the basic principles of inheritance, publishing his findings in 1866. Much of Mendel's success can be attributed to the seven characteristics of pea plants that he studied.

✓ CONCEPT CHECK 1

Which of the following factors did *not* contribute to Mendel's success in his study of heredity?

a. His use of the pea plant
b. His study of plant chromosomes
c. His adoption of an experimental approach
d. His use of mathematics

Genetic Terminology

Before we examine Mendel's crosses and the conclusions that he drew from them, a review of some terms commonly used in genetics will be helpful (**Table 3.1**). The term *gene* is a word that Mendel never knew. It was not coined until 1909, when Danish botanist Wilhelm Johannsen first used it.

TABLE 3.1 Summary of important genetic terms

Term	Definition
Gene	An inherited factor (region of DNA) that helps determine a characteristic
Allele	One of two or more alternative forms of a gene
Locus	A specific place on a chromosome occupied by an allele
Genotype	A set of alleles possessed by an individual organism
Homozygote	An individual organism possessing two of the same alleles at a locus
Heterozygote	An individual organism possessing two different alleles at a locus
Phenotype or trait	The appearance or manifestation of a characteristic
Characteristic or character	An attribute or a feature possessed by an organism

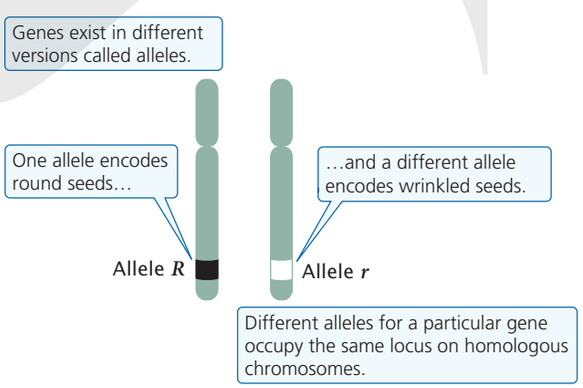
The definition of *gene* varies with the context of its use, so its definition will change as we explore different aspects of heredity. For our present use in the context of genetic crosses, we define a **gene** as an inherited factor that determines a characteristic.

Genes frequently come in different versions called **alleles** (**Figure 3.3**). In Mendel's crosses, seed shape was determined by a gene that exists as two different alleles: one allele encodes round seeds, and the other encodes wrinkled seeds. All alleles for any particular gene will be found at a specific place on a chromosome called the **locus**. (The plural of locus is **loci**; it's bad form in genetics—and incorrect—to speak of "locuses.") Thus, there is a specific place—a locus—on a chromosome in pea plants where the shape of seeds is determined. This locus may be occupied by an allele for round seeds or an allele for wrinkled seeds. We will use the term *allele* when referring to a specific version of a gene; we will use the term *gene* to refer more generally to any allele at a locus.

The **genotype** is the set of alleles that an individual organism possesses. A diploid organism with a genotype consisting of two identical alleles is **homozygous** at that locus. One that has a genotype consisting of two different alleles is **heterozygous** at the locus.

A **phenotype** is the manifestation or appearance of a characteristic. This term can refer to any type of characteristic—physical, physiological, biochemical, or behavioral. Thus, the condition of having round seeds is a phenotype, a body weight of 50 kilograms (50 kg) is a phenotype, and having sickle-cell anemia is a phenotype. In this book, the term *characteristic* or *character* refers to a general feature such as eye color; the term *trait* or *phenotype* refers to specific manifestations of that feature, such as blue or brown eyes.

A given phenotype arises from a genotype that develops within a particular environment. The genotype



3.3 At each locus, a diploid organism possesses two alleles located on different homologous chromosomes. The alleles identified here refer to alleles in pea plants studied by Mendel.

determines the potential for development; it sets certain limits, or boundaries, on that development. How the phenotype develops within those limits is determined by the effects of other genes and of environmental factors, and the balance between these effects varies from characteristic to characteristic. For some characteristics, differences between phenotypes are determined largely by differences in genotype. In Mendel's peas, for example, the genotype, not the environment, largely determined the shape of the seeds. For other characteristics, environmental differences are more important. The height reached by an oak tree at maturity is a phenotype that is strongly influenced by environmental factors, such as the availability of water, sunlight, and nutrients. Nevertheless, the tree's genotype imposes some limits on its height: an oak tree will never grow to be 300 meters (984 feet) tall, no matter how much sunlight, water, and fertilizer are provided. Thus, even the height of an oak tree is determined to some degree by genes. For many characteristics, both genes and environment are important in determining phenotypic differences.

An obvious but important point is that only the alleles of the genotype are inherited. Although the phenotype is determined, at least to some extent, by the genotype, organisms do not transmit their phenotypes to the next generation. The distinction between genotype and phenotype is one of the most important principles of modern genetics. The next section describes Mendel's careful observation of phenotypes through several generations of breeding experiments. These experiments allowed him to deduce not only the genotypes of individual pea plants but also the rules governing their inheritance.

CONCEPTS

Each phenotype results from a genotype developing within a specific environment. The alleles of the genotype, not the phenotype, are inherited.

✓ CONCEPT CHECK 2

What is the difference between a locus and an allele? What is the difference between a genotype and a phenotype?

3.2 Monohybrid Crosses Reveal the Principle of Segregation and the Concept of Dominance

Starting with 34 varieties of peas, Mendel spent two years selecting those varieties that he would use in his experiments. He verified that each variety was pure-breeding (homozygous for each of the traits that he chose to study) by growing the plants for two generations and confirming that all offspring were the same as their parents. He then carried out a number of crosses between the different varieties. Although peas normally self-fertilize (each plant mates with itself), Mendel conducted crosses between different plants by opening the buds before the anthers (male sex organs) were fully developed, removing the anthers, and then dusting the stigma (female sex organ) with pollen from a different plant's anthers (**Figure 3.4**).

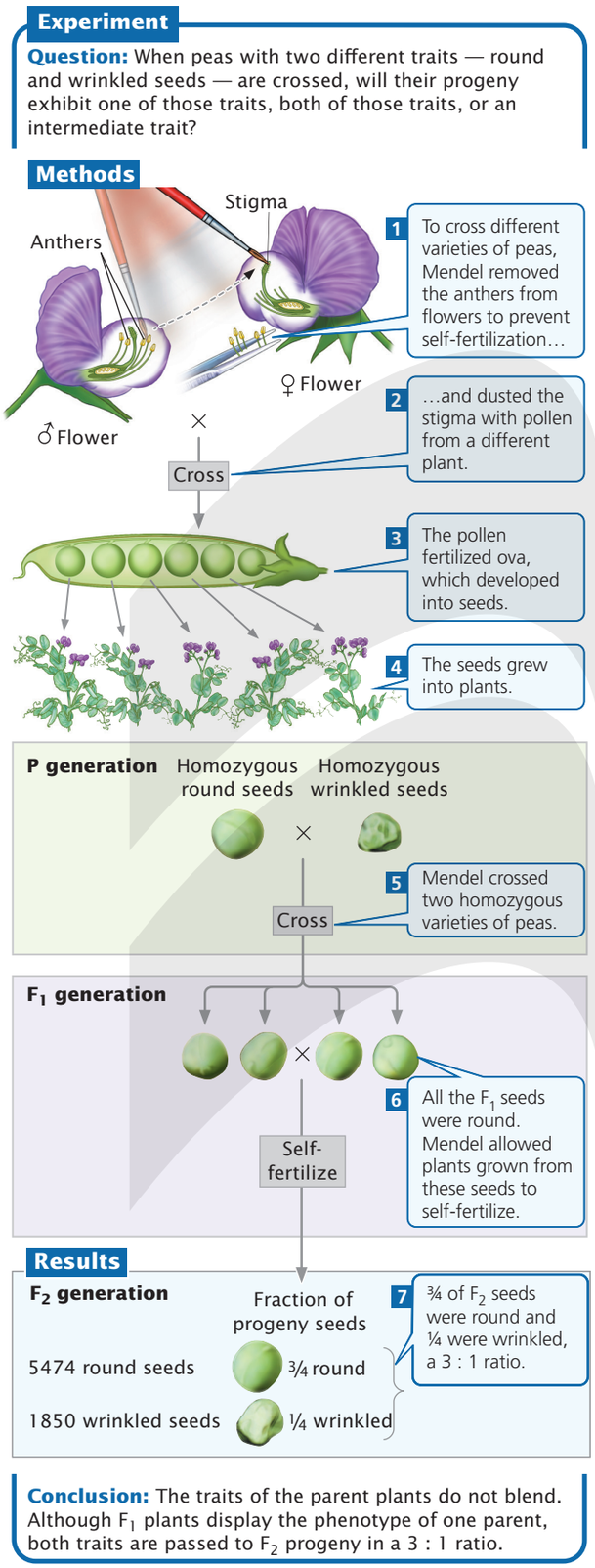
Mendel began by studying **monohybrid crosses**—crosses between parents that differed in a single characteristic. In one experiment, Mendel crossed a pea plant that was pure-breeding (homozygous) for round seeds with one that was pure-breeding for wrinkled seeds (see Figure 3.4). This first generation of a cross is called the **P (parental) generation**.

After crossing the two varieties in the P generation, Mendel observed the offspring that resulted from the cross. The seed shape phenotype develops as soon as the seed matures because seed traits are determined by the newly formed embryo within the seed. For characteristics associated with the plant itself, such as stem length, the phenotype doesn't develop until the plant grows from the seed; for these characteristics, Mendel had to wait until the following spring, plant the seeds, and then observe the phenotypes of the plants that germinated.

The offspring of the parents in the P generation are the **F_1 (first filial) generation**. When Mendel examined the F_1 generation of this cross, he found that they expressed only one of the phenotypes present in the parental generation: all the F_1 seeds were round. Mendel carried out 60 such crosses and always obtained this result. Furthermore, he conducted **reciprocal crosses**: in one cross, pollen (the male gamete) was taken from a plant with round seeds, and in its reciprocal cross, pollen was taken from a plant with wrinkled seeds. Reciprocal crosses gave the same result: all the F_1 seeds were round.

THINK-PAIR-SHARE Question 5

Mendel wasn't content with examining only the seeds arising from these monohybrid crosses, however. The following spring, he planted the F_1 seeds, cultivated the plants that germinated from them, and allowed the plants to self-fertilize, producing a second generation—the **F_2 (second filial) generation**. Both of the traits from the P generation emerged in the F_2 generation; Mendel counted 5474 round seeds and 1850 wrinkled seeds in the F_2 (see Figure 3.4). He noticed that the numbers of the round and wrinkled seeds constituted approximately a 3 : 1 ratio; that is, about $\frac{3}{4}$ of the F_2 seeds were round and $\frac{1}{4}$ were wrinkled. Mendel conducted monohybrid crosses for all seven of the characteristics that he studied in pea plants, and in all the crosses, he obtained the same result: all the F_1 resembled only one of the two parents, but both parental traits emerged in the F_2 in an approximate ratio of 3 : 1.



3.4 Mendel conducted monohybrid crosses.

What Monohybrid Crosses Reveal

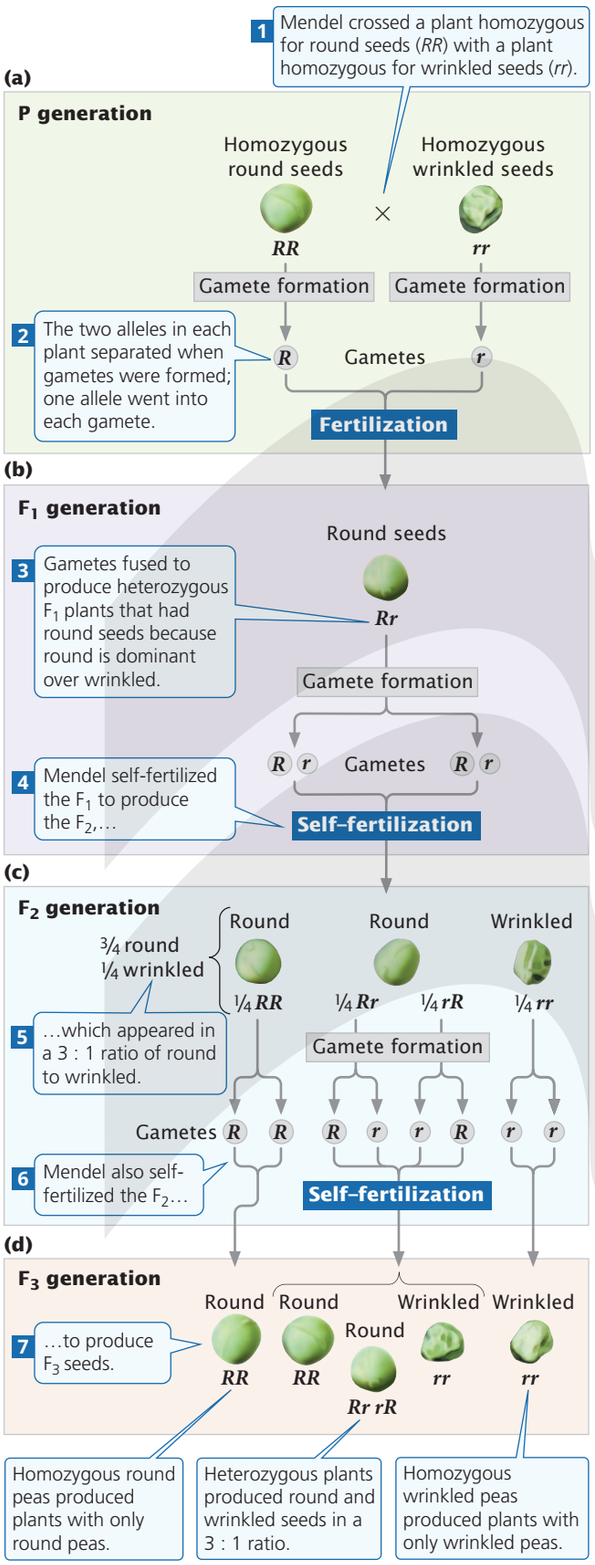
Mendel drew several conclusions from the results of his monohybrid crosses. First, he reasoned that although the F_1 plants display the phenotype of only one parent, they must inherit genetic factors from both parents because they transmit both parental phenotypes to the F_2 generation. The presence of both round and wrinkled seeds in the F_2 plants could be explained only if the F_1 plants possessed both round and wrinkled genetic factors that they had inherited from the P generation. He concluded that each plant must therefore possess two genetic factors encoding a characteristic.

The genetic factors (now called alleles) that Mendel discovered are, by convention, designated with letters: the allele for round seeds is usually represented by *R* and the allele for wrinkled seeds by *r*. The plants in the P generation of Mendel's cross possessed two identical alleles: *RR* in the round-seeded parent and *rr* in the wrinkled-seeded parent (**Figure 3.5a**).

The second conclusion that Mendel drew from his monohybrid crosses was that the two alleles in each plant separate when gametes are formed, and one allele goes into each gamete. When two gametes (one from each parent) fuse to produce a zygote, the allele from the male parent unites with the allele from the female parent to produce the genotype of the offspring. Thus, Mendel's F_1 plants inherited an *R* allele from the round-seeded plant and an *r* allele from the wrinkled-seeded plant (**Figure 3.5b**). However, only the trait encoded by the round allele (*R*) was observed in the F_1: all the F_1 progeny had round seeds. Those traits that appeared unchanged in the F_1 heterozygous offspring Mendel called **dominant**, and those traits that disappeared in the F_1 heterozygous offspring he called **recessive**. In plants, alleles for dominant traits are often symbolized with uppercase letters (e.g., *R*), while alleles for recessive traits are often symbolized with lowercase letters (e.g., *r*). When dominant and recessive alleles are present together, the effect of the recessive allele is not observed in the phenotype. This concept of dominance was the third important conclusion that Mendel derived from his monohybrid crosses.

Mendel's fourth conclusion was that the two alleles of an individual plant separate with equal probability into the gametes. When plants of the F_1 (with genotype *Rr*) produced gametes, half of the gametes received the *R* allele for round seeds and half received the *r* allele for wrinkled seeds. The gametes then paired randomly to produce the following genotypes in equal proportions among the F_2: *RR*, *Rr*, *rR*, and *rr* (**Figure 3.5c**). Because round (*R*) is dominant to wrinkled (*r*), there were three round-seeded progeny (*RR*, *Rr*, and *rR*) for every wrinkled-seeded progeny (*rr*) in the F_2. This 3 : 1 ratio of round-seeded to wrinkled-seeded progeny that Mendel observed in the F_2 could be obtained only if the two alleles of a genotype separated into the gametes with equal probability.

The conclusions that Mendel made about inheritance from his monohybrid crosses have been further developed



3.5 Mendel's monohybrid crosses revealed the principle of segregation and the concept of dominance.

TABLE 3.2 Comparison of the principles of segregation and independent assortment

Principle	Observation	Stage of meiosis*
Segregation (Mendel's first law)	1. Each individual organism possesses two alleles encoding a trait.	Before meiosis
	2. Alleles separate when gametes are formed.	Anaphase I
	3. Alleles separate in equal proportions.	Anaphase I
Independent assortment (Mendel's second law)	Alleles at different loci separate independently.	Anaphase I

*Assumes that no crossing over occurs. If crossing over takes place, then segregation and independent assortment may also occur in anaphase II of meiosis.

and formalized into the principle of segregation and the concept of dominance. The **principle of segregation (Mendel's first law)** (**Table 3.2**) states that each individual diploid organism possesses two alleles for any particular characteristic, one inherited from the maternal parent and one from the paternal parent. These two alleles segregate (separate) when gametes are formed, and one allele goes into each gamete. Furthermore, the two alleles segregate into gametes in equal proportions. The **concept of dominance** states that when two different alleles are present in a genotype, only the trait encoded by one of them—the dominant allele—is observed in the phenotype.

Mendel confirmed these principles by allowing his F_2 plants to self-fertilize and produce an F_3 generation. He found that the plants grown from the wrinkled seeds—those displaying the recessive trait (*rr*)—produced an F_3 in which all plants produced wrinkled seeds. Because his wrinkled-seeded plants were homozygous for wrinkled alleles (*rr*), only wrinkled alleles could be passed on to their progeny (**Figure 3.5d**).

The plants grown from round seeds—the dominant trait—fell into two types (see Figure 3.5c). With self-fertilization, about $\frac{2}{3}$ of these plants produced both round-seeded and wrinkled-seeded progeny in the F_3 generation. These plants were heterozygous (*Rr*), so they produced $\frac{1}{4}$ *RR* (round), $\frac{1}{2}$ *Rr* (round), and $\frac{1}{4}$ *rr* (wrinkled) progeny, giving a 3 : 1 ratio of round to wrinkled in the F_3. About $\frac{1}{3}$ of the plants grown from round seeds were of the second type; they produced only the round-seeded trait in the F_3. These plants were homozygous for the round allele (*RR*) and could thus produce only round-seeded offspring in the F_3 generation. Mendel planted the seeds obtained in the F_3 and carried these plants through three more rounds of self-fertilization. In each generation, $\frac{2}{3}$ of the round-seeded plants produced round and wrinkled offspring, whereas $\frac{1}{3}$ produced only round offspring. These results are entirely consistent with the principle of segregation.

CONCEPTS

The principle of segregation states that each individual organism possesses two alleles that can encode a characteristic. These alleles segregate when gametes are formed, and one allele goes into each gamete. The concept of dominance states that when the two alleles of a genotype are different, only the trait encoded by one of them—the "dominant" allele—is observed.

✓ CONCEPT CHECK 3

How did Mendel know that each of his pea plants carried two alleles encoding a characteristic?

The Molecular Nature of Alleles

What exactly is an allele, and how does it determine a phenotype? Although Mendel had no information about the physical nature of the genetic factors in his crosses, modern geneticists have now determined the molecular basis of those factors and how they encode a trait such as wrinkled peas.

Alleles, such as the *R* and *r* alleles that encode round and wrinkled peas, usually represent specific DNA sequences. The locus that determines whether a pea is round or wrinkled is a sequence of DNA on pea chromosome 5 that encodes a protein called starch-branching enzyme isoform I (SBEI). The *R* allele, which produces round seeds in pea plants, encodes a normal, functional form of the SBEI enzyme. This enzyme converts a linear form of starch into a highly branched form. The *r* allele, which encodes wrinkled seeds, is a different DNA sequence that contains a mutation or error; it encodes an inactive form of the enzyme that does not produce the branched form of starch and leads to the accumulation of sucrose within the *rr* seed (the pea). Because the *rr* seed contains a large amount of sucrose, the developing seed absorbs water and swells. Later, as the seed matures, it loses the excess water and afterward appears shriveled or wrinkled. The *r* allele for wrinkled seeds is recessive because the presence of a single *R* allele in the heterozygote allows the plant to synthesize enough SBEI enzyme to produce branched starch and therefore round seeds.

Research has revealed that the *r* allele contains an extra 800 base pairs of DNA, which disrupt the normal coding sequence of the gene. The extra DNA appears to have come from a transposable element—a type of DNA sequence that has the ability to move from one location in the genome to another—which we will discuss further in Chapter 13.

Predicting the Outcomes of Genetic Crosses

One of Mendel's goals in conducting his experiments with pea plants was to develop a way to predict the outcomes of crosses between plants with different phenotypes. In this section, you will first learn a simple shorthand method for predicting outcomes of genetic crosses (the Punnett square), and then you will learn how to use probability to predict the results of crosses.

CONNECTING CONCEPTS

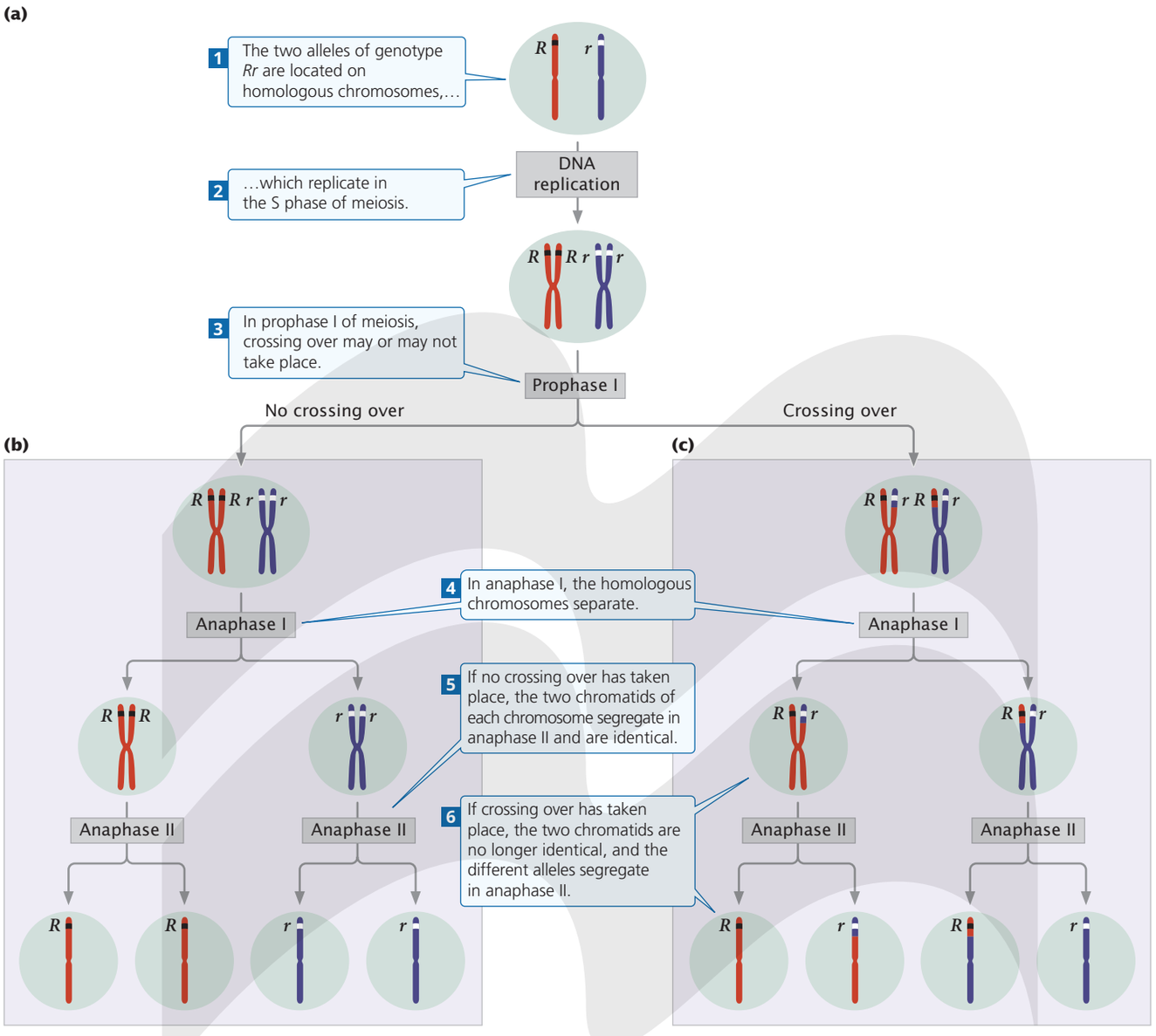
Relating Genetic Crosses to Meiosis

We have now seen how the results of monohybrid crosses are explained by Mendel's principle of segregation. Many students find that they enjoy working genetic crosses but are frustrated by the abstract nature of the symbols. Perhaps you feel the same at this point. You may be asking, "What do these symbols really represent? What does the genotype *RR* mean in regard to the biology of the organism?" The answers to these questions lie in relating the abstract symbols of crosses to the structure and behavior of chromosomes, the repositories of genetic information (see Chapter 2).

In 1900, when Mendel's work was rediscovered and biologists began to apply his principles of heredity, the relation between genes and chromosomes was still unclear. The theory that genes are located on chromosomes (the **chromosome theory of heredity**) was developed in the early 1900s by Walter Sutton, then a graduate student at Columbia University. Through the careful study of meiosis in insects, Sutton documented the fact that each homologous pair of chromosomes consists of one maternal chromosome and one paternal chromosome. Showing that these pairs segregate independently into gametes in meiosis, he concluded that this process is the biological basis for Mendel's principles of heredity. German cytologist and embryologist Theodor Boveri came to similar conclusions at about the same time.

The symbols used in genetic crosses, such as *R* and *r*, are just shorthand notations for particular sequences of DNA in the chromosomes that encode particular phenotypes. The two alleles of a genotype are found on different but homologous chromosomes. One chromosome of each homologous pair is inherited from the mother, and the other is inherited from the father. In the S phase of meiotic interphase, each chromosome replicates, producing two copies of each allele, one on each chromatid (**Figure 3.6a**). The homologous chromosomes segregate in anaphase I, thereby separating the two different alleles (**Figure 3.6b**). This chromosome segregation is the basis of the principle of segregation. In anaphase II of meiosis, the two chromatids of each replicated chromosome separate, so each gamete resulting from meiosis carries only a single allele at each locus, as Mendel's principle of segregation predicts.

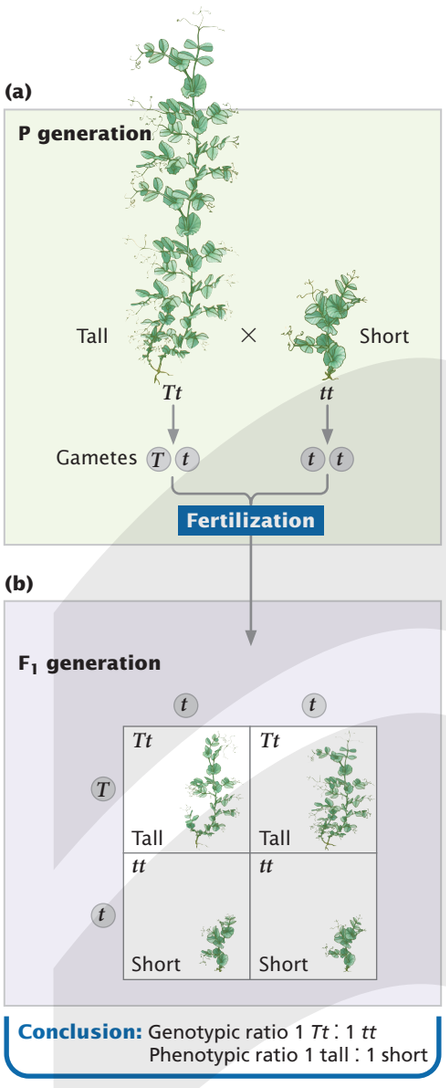
If crossing over has taken place in prophase I of meiosis, then the two chromatids of each replicated chromosome are no longer identical, and the segregation of different alleles takes place at anaphase I and anaphase II (**Figure 3.6c**). However, Mendel didn't know anything about chromosomes; he formulated his principles of heredity entirely on the basis of the results of the crosses that he carried out. Nevertheless, we should not forget that these principles work because they are based on the behavior of actual chromosomes in meiosis. TRY PROBLEM 28



3.6 Segregation results from the separation of homologous chromosomes in meiosis.

The Punnett Square The Punnett square was developed by the English geneticist Reginald C. Punnett in 1917. To illustrate the Punnett square, let's examine another cross carried out by Mendel. By crossing two varieties of pea plants that differed in height, Mendel established that tall (*T*) was dominant to short (*t*). He tested his theory concerning the inheritance of dominant traits by crossing an F_1 tall plant that was heterozygous (*Tt*) with the short homozygous parental variety (*tt*). This type of cross, between an F_1 genotype and either of the parental genotypes, is called a **backcross**.

To predict the types of offspring that will result from this backcross, we must first determine which gametes will be produced by each parent (**Figure 3.7a**). The principle of segregation tells us that the two alleles in each parent separate and that one allele passes to each gamete. All gametes from the homozygous *tt* short plant will receive a single short (*t*) allele. The tall plant in this cross is heterozygous (*Tt*); thus, 50% of its gametes will receive a tall allele (*T*), and the other 50% will receive a short allele (*t*).



3.7 The Punnett square can be used to determine the results of a genetic cross.

A **Punnett square** is constructed by drawing a grid, listing the gametes produced by one parent along the upper edge, and listing the gametes produced by the other parent down the left side (**Figure 3.7b**). Each cell (that is, each block within the Punnett square) is then filled in with an allele from each of the corresponding gametes, generating the genotype of the progeny produced by the fusion of those gametes. In the upper left-hand cell of the Punnett square in Figure 3.7b, a gamete containing *T* from the tall plant unites with a gamete containing *t* from the short plant, giving the genotype of the progeny (*Tt*). It is useful to write the phenotype expressed by each genotype; here, the progeny will be tall because the tall allele is dominant to the short allele. This process is repeated for all the cells in the Punnett square.

By simply counting, we can determine the types of progeny produced and their ratios. In Figure 3.7b, two cells contain tall (*Tt*) progeny and two cells contain short (*tt*) progeny, so the genotypic ratio expected for this cross is 2 *Tt* to 2 *tt* (a 1 : 1 ratio). Another way to express this result is to say that we expect ½ of the progeny to have genotype *Tt* (the tall phenotype) and ½ of the progeny to have genotype *tt* (the short phenotype).

In this cross, the genotypic ratio and the phenotypic ratio are the same, but this outcome need not be the case for all crosses. Try completing a Punnett square for the cross in which the F_1 round-seeded plants in Figure 3.5b undergo self-fertilization (you should obtain a phenotypic ratio of 3 round to 1 wrinkled and a genotypic ratio of 1 *RR* to 2 *Rr* to 1 *rr*).

CONCEPTS

The Punnett square is a shorthand method of predicting the genotypic and phenotypic ratios of progeny from a genetic cross.

✓ CONCEPT CHECK 4

If the F_1 plant depicted in Figure 3.5b is backcrossed to the parent with round seeds, what proportion of the progeny will have wrinkled seeds? (Use a Punnett square.)

a. ¾ c. ¼
b. ½ d. 0

Probability as a Tool of Genetics Another method for determining the outcome of a genetic cross is to use the rules of probability, as Mendel did with his crosses. **Probability** expresses the likelihood of the occurrence of a particular event. It is the number of times that a particular event takes place, divided by the number of all possible outcomes. For example, a deck of 52 cards contains only one king of hearts. The probability of drawing one card from the deck at random and obtaining the king of hearts is $\frac{1}{52}$ because there is only one card that is the king of hearts (one event) and there are 52 cards that can be drawn from the deck (52 possible outcomes). The probability of drawing a card and obtaining an ace is $\frac{4}{52}$ because there are four cards that are aces (four events) and 52 cards (possible outcomes). Probability can be expressed either as a fraction ($\frac{4}{52}$ in this case) or as a decimal number (0.077 in this case).

The probability of a particular event may be determined by knowing something about *how* or *how often* the event takes place. We know, for example, that the probability of rolling a six-sided die and getting a four is ⅙ because the die has six sides and any one side is equally likely to end up on top. So, in this case, understanding the nature of the

event—the shape of the thrown die—allows us to determine its probability. In other cases, we determine the probability of an event by making a large number of observations. When a weather forecaster says that there is a 40% chance of rain on a particular day, this probability was obtained by observing a large number of days with similar atmospheric conditions and finding that it rains on 40% of those days. In this case, the probability has been determined empirically (by observation).

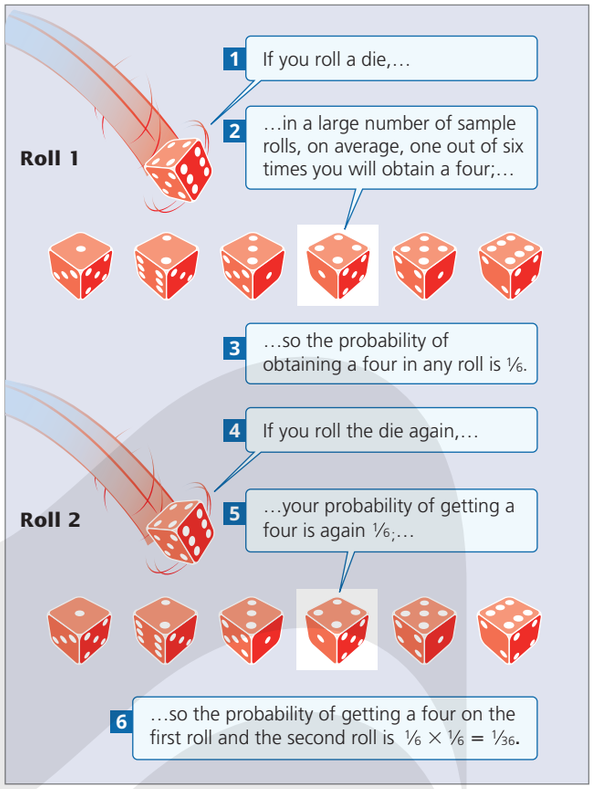
The Multiplication Rule Two rules of probability are useful for predicting the ratios of offspring from genetic crosses. The first is the **multiplication rule**, which states that the probability of two or more independent events taking place together is calculated by multiplying their independent probabilities.

To illustrate the use of the multiplication rule, let's again consider the roll of a die. The probability of rolling one die and obtaining a four is $\frac{1}{6}$. To calculate the probability of rolling a die twice and obtaining two fours, we can apply the multiplication rule. The probability of obtaining a four on the first roll is $\frac{1}{6}$ and the probability of obtaining a four on the second roll is $\frac{1}{6}$, so the probability of rolling a four on both is $\frac{1}{6} \times \frac{1}{6} = \frac{1}{36}$ (**Figure 3.8a**). The key indicator for applying the multiplication rule is the word *and*; in the example just considered, we wanted to know the probability of obtaining a four on the first roll *and* a four on the second roll. (It may have been some time since you worked with fractions. If you need a review, visit the *Genetics Essentials* catalog page to access the "Working with Fractions" resource.)

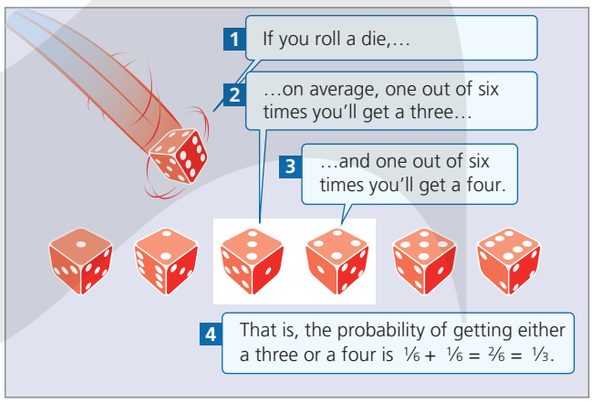
For the multiplication rule to be valid, the events whose joint probability is being calculated must be independent—the outcome of one event must not influence the outcome of the other. For example, the number that comes up on one roll of the die has no influence on the number that comes up on the next roll, so these events are independent. However, if we wanted to know the probability of being hit on the head with a hammer and going to the hospital on the same day, we could not simply apply the multiplication rule and multiply the two probabilities together because the two events are not independent—being hit on the head with a hammer certainly influences the probability of going to the hospital.

The Addition Rule The second rule of probability frequently used in genetics is the **addition rule**, which states that the probability of any of two or more mutually exclusive events taking place is calculated by adding the probabilities of the events. Let's look at this rule in concrete terms. To obtain the probability of throwing a die once and rolling *either* a three *or* a four, we would use the addition rule, adding the probability of obtaining a three ($\frac{1}{6}$) to the probability of obtaining a four (again, $\frac{1}{6}$), or $\frac{1}{6} + \frac{1}{6} = \frac{2}{6} = \frac{1}{3}$

(a) The multiplication rule



(b) The addition rule



3.8 The multiplication and addition rules can be used to determine the probability of combinations of events.

(**Figure 3.8b**). The key indicators for applying the addition rule are the words *either* and *or*.

For the addition rule to be valid, the events whose probability is being calculated must be mutually exclusive, meaning that one event excludes the possibility of the other. For example, you cannot throw a single die just once and obtain both a three and a four because only one side of the die can be on top. These events are mutually exclusive.

CONCEPTS

The multiplication rule states that the probability of two or more independent events taking place together is calculated by multiplying their independent probabilities. The addition rule states that the probability that any one of two or more mutually exclusive events taking place is calculated by adding their probabilities.

✓ CONCEPT CHECK 5

If the probability of being blood type A is $\frac{1}{8}$ and the probability of being blood type O is $\frac{1}{2}$, what is the probability of being either blood type A or blood type O?

a. $\frac{5}{8}$ c. $\frac{1}{10}$
b. $\frac{1}{2}$ d. $\frac{1}{16}$

Applying Probability to Genetic Crosses The multiplication and addition rules of probability can be used in place of the Punnett square to predict the ratios of progeny expected from a genetic cross. Let's first consider a cross between two pea plants heterozygous for the locus that determines height, $Tt \times Tt$. Half of the gametes produced by each plant have a *T* allele, and the other half have a *t* allele, so the probability for each type of gamete is $\frac{1}{2}$.

The gametes from the two parents can combine in four different ways to produce offspring. Using the multiplication rule, we can determine the probability of each possible combination. To calculate the probability of obtaining *TT* progeny, for example, we multiply the probability of receiving a *T* allele from the first parent ($\frac{1}{2}$) by the probability of receiving a *T* allele from the second parent ($\frac{1}{2}$). The multiplication rule should be used here because we need the probability of receiving a *T* allele from the first parent *and* a *T* allele from the second parent—two independent events. The four types of progeny from this cross and their associated probabilities are

TT	(*T* gamete and *T* gamete)	$\frac{1}{2} \times \frac{1}{2} = \frac{1}{4}$	tall
Tt	(*T* gamete and *t* gamete)	$\frac{1}{2} \times \frac{1}{2} = \frac{1}{4}$	tall
tT	(*t* gamete and *T* gamete)	$\frac{1}{2} \times \frac{1}{2} = \frac{1}{4}$	tall
tt	(*t* gamete and *t* gamete)	$\frac{1}{2} \times \frac{1}{2} = \frac{1}{4}$	short

Notice that there are two ways for heterozygous progeny to be produced: a heterozygote can either receive a *T* allele from the first parent and a *t* allele from the second, or it can receive a *t* allele from the first parent and a *T* allele from the second.

After determining the probability of obtaining each progeny genotype, we can use the addition rule to determine the overall phenotypic ratios. Because of dominance, a tall plant can have genotype *TT*, *Tt*, or *tT;* thus using the addition rule, we find the probability of tall progeny to be $\frac{1}{4} + \frac{1}{4} + \frac{1}{4} = \frac{3}{4}$. Because only one genotype (*tt*) encodes the short phenotype, the probability of short progeny is simply $\frac{1}{4}$.

Two methods have now been introduced to work genetic crosses: the Punnett square and the probability method. At this point, you may be asking, "Why bother with probability rules and calculations? The Punnett square is easier to understand and just as quick." This is true for simple monohybrid crosses. For tackling more complex crosses that assess genes at two or more loci, however, the probability method is both clearer and quicker than the Punnett square.

Conditional Probability Thus far, we have used probability to predict the chances of producing certain types of progeny given only the genotypes of the parents. Sometimes, however, we have additional information that modifies, or conditions, the probability, a situation termed **conditional probability**. For example, assume that we cross two heterozygous pea plants ($Tt \times Tt$) and obtain a tall progeny plant. What is the probability that this tall plant is heterozygous (*Tt*)? You might assume that the probability would be $\frac{1}{2}$, the probability of obtaining heterozygous progeny in a cross between two heterozygotes. In this case, however, we have some additional information—the phenotype of the progeny plant—which modifies that probability. When two heterozygous individuals are crossed, we expect $\frac{1}{4}$ *TT*, $\frac{1}{2}$ *Tt*, and $\frac{1}{4}$ *tt* progeny. We know that the plant in question is tall, so we can eliminate the possibility that it has genotype *tt*. Tall progeny must be either genotype *TT* or genotype *Tt*, and in a cross between two heterozygotes, these genotypes occur in a 1 : 2 ratio. Therefore, the probability that a tall progeny plant is heterozygous (*Tt*) is two out of three, or $\frac{2}{3}$.

TRY PROBLEMS 24 & 25

THINK-PAIR-SHARE Question 6

The Testcross

A useful tool for analyzing genetic crosses is the **testcross**, in which one individual of unknown genotype is crossed with another individual with a homozygous recessive genotype for the trait in question. Figure 3.7 illustrates a testcross (in this case, it is also a backcross). A testcross tests, or reveals, the genotype of the first individual.

Suppose you were given a tall pea plant with no information about its parents. Because tallness is a dominant trait in peas, your plant could be either homozygous (*TT*) or heterozygous (*Tt*) for the dominant allele, but you would not know which. You could determine its genotype by performing a testcross. If the plant were homozygous (*TT*), a testcross would produce all tall progeny ($TT \times tt \rightarrow$ all *Tt*); if the plant were heterozygous (*Tt*), approximately half of the progeny would be tall and approximately half would be short ($Tt \times tt \rightarrow \frac{1}{2}$ *Tt* and $\frac{1}{2}$ *tt*). When a testcross is performed, any recessive allele in the unknown genotype will be expressed in the progeny because it will be paired with a recessive allele from the homozygous recessive parent.

TRY PROBLEM 19

CONCEPTS

A testcross is a cross between an individual with an unknown genotype and one with a homozygous recessive genotype. The outcome of the testcross can reveal the unknown genotype.

Genetic Symbols

As we have seen, genetic crosses are usually depicted with symbols that designate the different alleles. The symbols used for alleles are usually determined by the community of geneticists who work on a particular organism; therefore, there is no universal system for designating symbols. In plants, lowercase letters are often used to designate recessive alleles and uppercase letters to designate dominant alleles. Two or three letters may be used for a single allele: the recessive allele for heart-shaped leaves in cucumbers is designated *hl*, and the recessive allele for abnormal sperm-head shape in mice is designated *azh*.

In animals, the most common allele for a characteristic—called the **wild type** because it is the allele usually found in the wild—is often symbolized by one or more letters and a plus sign (+). The letter or letters chosen are usually based on a mutant (less common) phenotype. For example, the recessive allele that encodes yellow eyes in the Oriental fruit fly is represented by *ye*, whereas the allele for wild-type eye color is represented by ye^+. At times, the letters for the wild-type allele are dropped and the allele is represented simply by a plus sign.

Gene names and abbreviations are usually italicized. Superscripts and subscripts are sometimes added to distinguish between genes: Lfr_1 and Lfr_2 represent dominant mutant alleles at different loci that produce lacerate leaf margins in opium poppies; El^R represents an allele in goats that restricts the length of the ears. A slash may be used to distinguish the two alleles present in an individual genotype. For example, the genotype of a goat that is heterozygous for restricted ears might be El^+/El^R, or simply $+/El^R$. If genotypes at more than one locus are presented together, a space separates the genotypes. For example, a goat heterozygous for a pair of alleles that produces restricted ears and heterozygous for another pair of alleles that produces goiter can be designated El^+/El^R *G*/*g*. Sometimes it is useful to designate the possibility of several genotypes. An underline in a genotype, such as *A*_, indicates that any allele is possible. In this case, *A*_ might include both *AA* and *Aa* genotypes.

CONNECTING CONCEPTS

Ratios in Simple Crosses

Now that we have had some experience with genetic crosses, let's review the ratios that appear in the progeny of simple crosses in which a single locus is under consideration and one of the traits exhibits dominance. Understanding these ratios and the parental genotypes that produce them will enable you to work simple genetic crosses quickly, without resorting to the Punnett square. Later in this chapter, we will use these ratios to work more complicated crosses that include several loci.

TABLE 3.3 Phenotypic ratios for simple genetic crosses (crosses for a single locus)

Phenotypic ratio	Genotypes of parents	Genotypes of progeny
3 : 1	*Aa* × *Aa*	$\frac{3}{4}$ *A*_ : $\frac{1}{4}$ *aa*
1 : 1	*Aa* × *aa*	$\frac{1}{2}$ *Aa* : $\frac{1}{2}$ *aa*
Uniform progeny	*AA* × *AA*	All *AA*
	aa × *aa*	All *aa*
	AA × *aa*	All *Aa*
	AA × *Aa*	All *A*_

There are only three phenotypic ratios to understand (**Table 3.3**). The 3 : 1 ratio arises in a simple genetic cross when both of the parents are heterozygous for a dominant trait (*Aa* × *Aa*). The second phenotypic ratio is the 1 : 1 ratio, which results from the mating of a heterozygous parent and a homozygous parent. To obtain this 1 : 1 ratio, the homozygous parent in this cross (*Aa* × *aa*) must carry two recessive alleles to produce progeny of which half display the recessive trait. A cross between a homozygous dominant parent and a heterozygous parent (*AA* × *Aa*) produces progeny displaying only the dominant trait.

The third phenotypic ratio is not really a ratio: all the progeny have the same phenotype. Several combinations of parents can produce this outcome (see Table 3.3). A cross between any two homozygous parents—either between two parents of the same homozygous genotype (*AA* × *AA* or *aa* × *aa*) or between two parents with different homozygous genotypes (*AA* × *aa*)—produces progeny all having the same phenotype. Progeny of a single phenotype can also result from a cross between a homozygous dominant parent and a heterozygote (*AA* × *Aa*).

If we are interested in the ratios of genotypes instead of phenotypes, there are again only three outcomes to remember (**Table 3.4**): the 1 : 2 : 1 ratio, produced by a cross between two heterozygotes; the 1 : 1 ratio, produced by a cross between a heterozygote and a homozygote; and the uniform progeny produced by a cross between two homozygotes. These simple phenotypic and genotypic ratios and the parental genotypes that produce them provide the key to understanding crosses for a single locus and, as you will see in the next section, for multiple loci.

TABLE 3.4 Genotypic ratios for simple genetic crosses (crosses for a single locus)

Genotypic ratio	Genotypes of parents	Genotypes of progeny
1 : 2 : 1	*Aa* × *Aa*	$\frac{1}{4}$ *AA* : $\frac{1}{2}$ *Aa* : $\frac{1}{4}$ *aa*
1 : 1	*Aa* × *aa*	$\frac{1}{2}$ *Aa* : $\frac{1}{2}$ *aa*
	Aa × *AA*	$\frac{1}{2}$ *Aa* : $\frac{1}{2}$ *AA*
Uniform progeny	*AA* × *AA*	All *AA*
	aa × *aa*	All *aa*
	AA × *aa*	All *Aa*

3.3 Dihybrid Crosses Reveal the Principle of Independent Assortment

We now extend Mendel's principle of segregation to some more complex crosses that include alleles at multiple loci. Understanding the nature of these crosses will require an additional principle: the principle of independent assortment.

Dihybrid Crosses

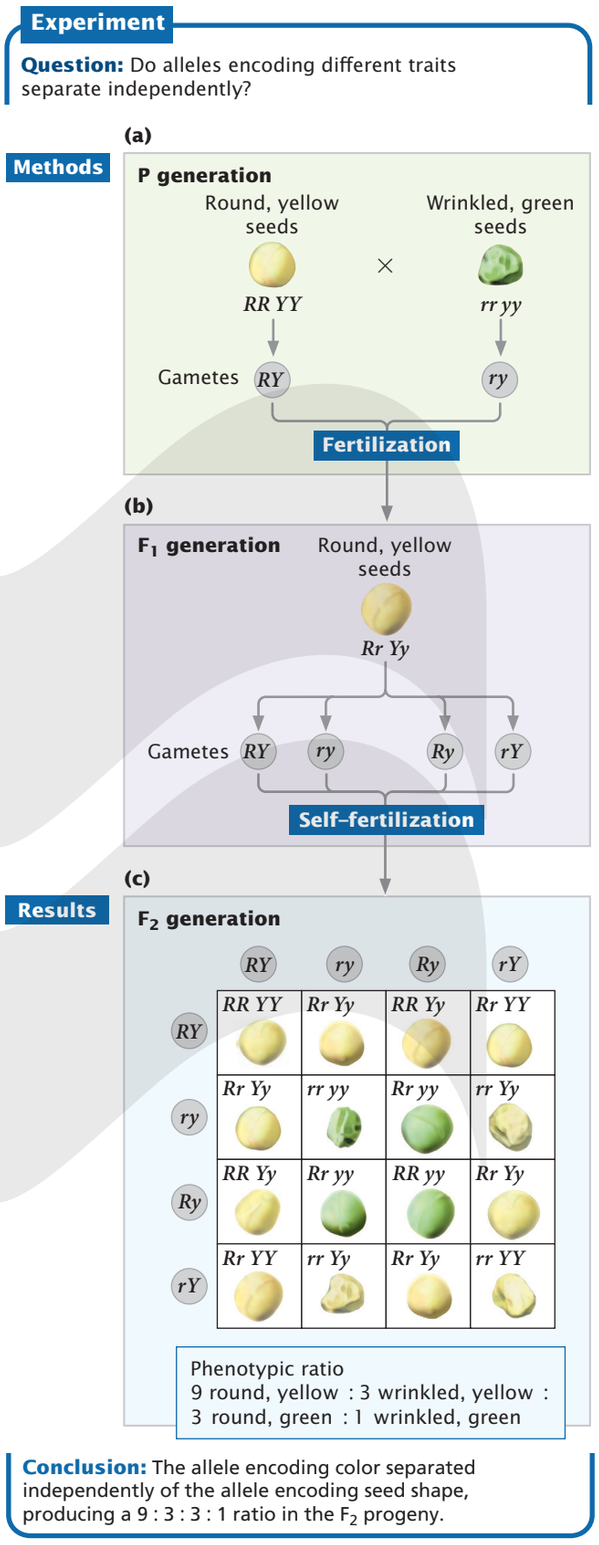
In addition to his work on monohybrid crosses, Mendel crossed varieties of peas that differed in *two* characteristics—that is, he performed **dihybrid crosses**. For example, he crossed one homozygous variety that had seeds that were round and yellow with another homozygous variety that had seeds that were wrinkled and green. The seeds of all the F_1 progeny were round and yellow. He then allowed the F_1 to self-fertilize and obtained the following progeny in the F_2: 315 round, yellow seeds; 101 wrinkled, yellow seeds; 108 round, green seeds; and 32 wrinkled, green seeds. Mendel recognized that these traits appeared in a ratio of approximately 9 : 3 : 3 : 1; that is, $\frac{9}{16}$ of the progeny were round and yellow, $\frac{3}{16}$ were wrinkled and yellow, $\frac{3}{16}$ were round and green, and $\frac{1}{16}$ were wrinkled and green.

The Principle of Independent Assortment

Mendel carried out a number of dihybrid crosses for pairs of characteristics and always obtained a 9 : 3 : 3 : 1 ratio in the F_2. This ratio makes perfect sense in regard to the principle of segregation and the concept of dominance if we add a third principle, which Mendel recognized in his dihybrid crosses: the **principle of independent assortment (Mendel's second law)**. This principle states that alleles at different loci separate independently of one another (see Table 3.2).

A common mistake is to think that the principle of segregation and the principle of independent assortment refer to two different processes. The principle of independent assortment is really an extension of the principle of segregation. The principle of segregation states that the two alleles at a locus separate when gametes are formed; the principle of independent assortment states that when these two alleles separate, their separation is independent of the separation of alleles at *other* loci.

Let's see how the principle of independent assortment explains the results that Mendel obtained in the dihybrid cross previously described. Each pea plant possesses two alleles encoding each characteristic, so the parent plants must have had genotypes *RR YY* and *rr yy* (**Figure 3.9a**). The principle of segregation tells us that the alleles at each locus separate, and that one allele for each locus passes into



3.9 Mendel's dihybrid crosses revealed the principle of independent assortment.

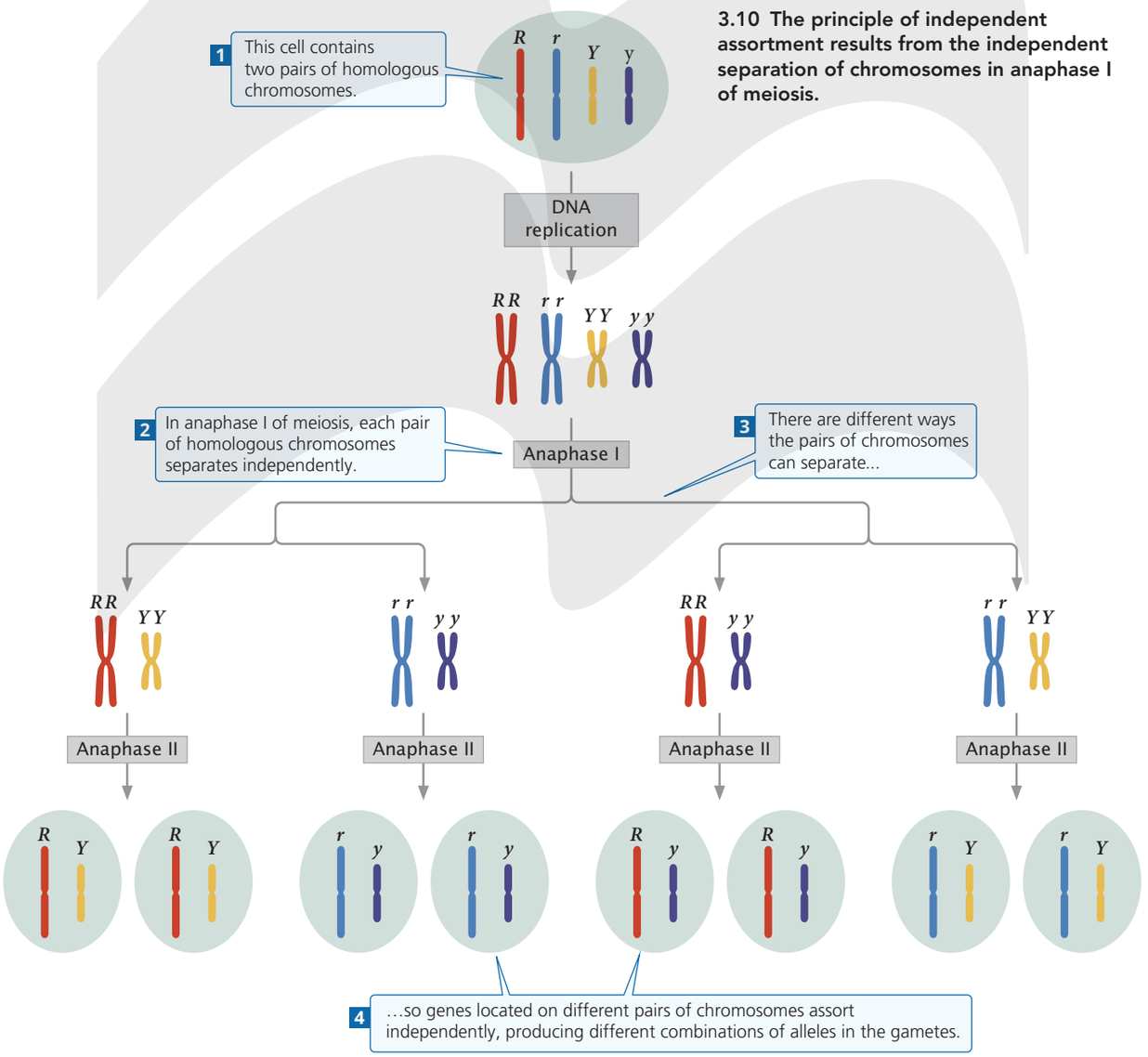
each gamete. The gametes produced by the round, yellow parent therefore contained alleles *RY*, whereas the gametes produced by the wrinkled, green parent contained alleles *ry*. These two types of gametes united to produce the F_1, all with genotype *Rr Yy*. Because round is dominant to wrinkled and yellow is dominant to green, the phenotype of the F_1 was round and yellow.

When Mendel allowed the F_1 plants to self-fertilize to produce the F_2, the alleles at each locus separated, and one of those alleles passed into each gamete. This event is where the principle of independent assortment becomes important. The two pairs of alleles can separate in two ways: (1) *R* separates with *Y*, and *r* separates with *y* to produce gametes *RY* and *ry*, or (2) *R* separates with *y*, and *r* separates with *Y* to produce gametes *Ry* and *rY*. The principle of independent assortment tells us that the alleles at each locus separate independently; thus, both kinds of separation take place with equal frequency, and all four types of gametes (*RY*, *ry*, *Ry*, and *rY*) are produced in equal proportions (**Figure 3.9b**). When these four types of gametes are combined to produce the F_2 generation, the progeny consist of $^9/_{16}$ round and yellow, $^3/_{16}$ wrinkled and yellow, $^3/_{16}$ round and green, and $^1/_{16}$ wrinkled and green, resulting in a 9 : 3 : 3 : 1 phenotypic ratio (**Figure 3.9c**).

THINK-PAIR-SHARE Question 7

Relating the Principle of Independent Assortment to Meiosis

An important qualification of the principle of independent assortment is that it applies to characteristics encoded by loci located on different chromosomes. Like the principle of segregation, it is based wholly on the behavior of chromosomes in meiosis. Each pair of homologous chromosomes separates independently of all other pairs in anaphase I of meiosis (**Figure 3.10**), so genes located on different pairs



3.10 The principle of independent assortment results from the independent separation of chromosomes in anaphase I of meiosis.

CONCEPTS

The principle of independent assortment states that genes encoding different characteristics separate independently of one another when gametes are formed, owing to the independent separation of homologous pairs of chromosomes in meiosis. Genes located close together on the same chromosome, however, do not assort independently.

✓ CONCEPT CHECK 6

How are the principles of segregation and independent assortment related, and how are they different?

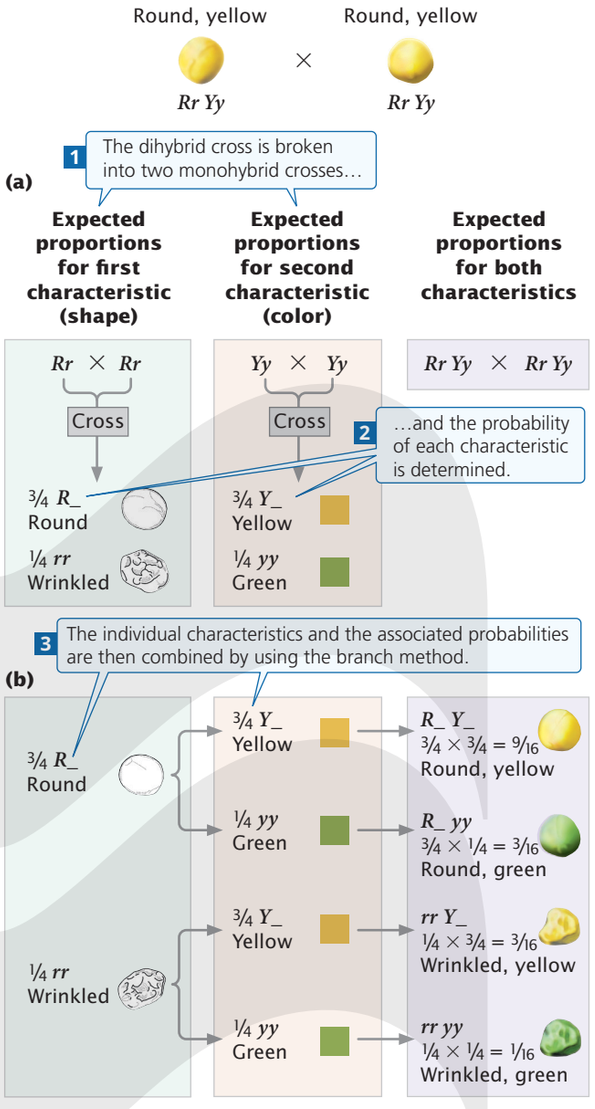
of homologs will assort independently. Genes that happen to be located on the same chromosome will travel together during anaphase I of meiosis and will arrive at the same destination—within the same gamete (unless crossing over takes place). So genes located on the same chromosome do not assort independently (unless they are located sufficiently far apart that crossing over takes place in every meiotic division, a situation that will be discussed fully in Section 5.2).

Applying Probability and the Branch Diagram to Dihybrid Crosses

When the genes at two loci separate independently, a dihybrid cross can be understood as two monohybrid crosses. Let's examine Mendel's dihybrid cross (*Rr Yy* × *Rr Yy*) by considering each characteristic separately (**Figure 3.11a**). If we consider only the shape of the seeds, the cross was *Rr* × *Rr*, which yields a 3 : 1 phenotypic ratio (¾ round and ¼ wrinkled progeny; see Table 3.3). Next, we consider the other characteristic, the color of the seeds. The cross was *Yy* × *Yy*, which produces a 3 : 1 phenotypic ratio (¾ yellow and ¼ green progeny).

We can now combine these monohybrid ratios by using the multiplication rule to obtain the proportion of progeny with different combinations of seed shape and color. The proportion of progeny with round and yellow seeds is ¾ (the probability of round) × ¾ (the probability of yellow) = 9/16. The proportion of progeny with round and green seeds is ¾ × ¼ = 3/16, the proportion of progeny with wrinkled and yellow seeds is ¼ × ¾ = 3/16, and the proportion of progeny with wrinkled and green seeds is ¼ × ¼ = 1/16.

Branch diagrams are a convenient way of organizing all the combinations of characteristics in the progeny of a cross (**Figure 3.11b**). In the first column, list the proportions of the phenotypes for one characteristic (here, seed shape: ¾ round and ¼ wrinkled). In the second column, list the proportions of the phenotypes for the next characteristic (seed color:



3.11 A branch diagram can be used to determine the phenotypes and expected proportions of offspring from a dihybrid cross (*Rr Yy* × *Rr Yy*).

¾ yellow and ¼ green) twice, next to each of the phenotypes in the first column; write "¾ yellow" and "¼ green" next to the round phenotype and again next to the wrinkled phenotype. Check to make sure that the proportions for each characteristic sum to one (i.e., ¾ yellow and ¼ green = 1.0). Now, draw lines between the phenotypes in the first column and each of the phenotypes in the second column. Follow each branch of the diagram, multiplying the probabilities for each trait along that branch. One branch leads from round to yellow, yielding round and yellow progeny. Another branch leads from round to green, yielding round and green progeny, and so forth. We calculate the probability of progeny

with a particular combination of traits by using the multiplication rule: the probability of round (¾) and yellow (¾) seeds is ¾ × ¾ = 9/16. The advantage of the branch diagram is that it helps us keep track of all the potential combinations of traits that may appear in the progeny. It can be used to determine phenotypic or genotypic ratios for any number of characteristics.

Using probability is much faster than using the Punnett square for crosses that include multiple loci. Genotypic and phenotypic ratios can be quickly worked out by combining, using the multiplication rule, the simple ratios in Tables 3.3 and 3.4. The probability method is particularly efficient if we need the probability of only a *particular* phenotype or genotype among the progeny of a cross. Suppose that we need to know the probability of obtaining the genotype *Rr yy* in the F_2 of the dihybrid cross in Figure 3.9. The probability of obtaining the *Rr* genotype in a cross of *Rr* × *Rr* is ½, and that of obtaining the *yy* genotype in a cross of *Yy* × *Yy* is ¼ (see Table 3.4). Using the multiplication rule, we find the probability of *Rr yy* to be ½ × ¼ = 1/8.

To illustrate the advantage of the probability method, consider the cross *Aa Bb cc Dd Ee* × *Aa Bb Cc dd Ee*. Suppose that we want to know the probability of obtaining offspring with the genotype *aa bb cc dd ee*. If we use a Punnett square to determine this probability, we might be working on the solution for months. However, we can quickly figure the probability of obtaining this one genotype by breaking this cross into a series of single-locus crosses.

Progeny cross	Genotype	Probability
Aa × *Aa*	*aa*	¼
Bb × *Bb*	*bb*	¼
cc × *Cc*	*cc*	½
Dd × *dd*	*dd*	½
Ee × *Ee*	*ee*	¼

The probability of an offspring from this cross having genotype *aa bb cc dd ee* is now easily obtained by using the multiplication rule: ¼ × ¼ × ½ × ½ × ¼ = 1/256. This calculation assumes that the genes at these five loci all assort independently.

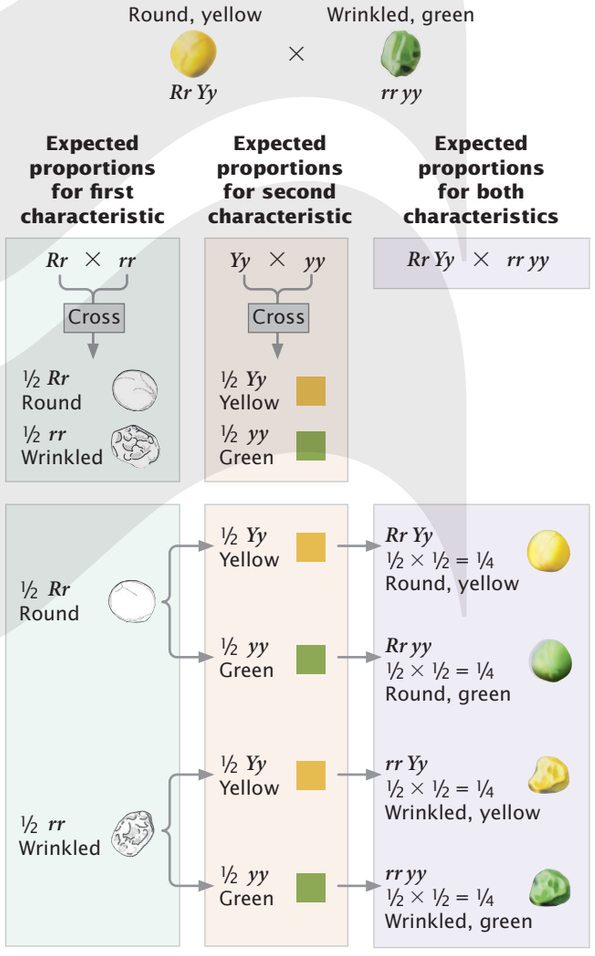
CONCEPTS

A cross including several characteristics can be worked by breaking it down into single-locus crosses and using the multiplication rule to determine the proportions of combinations of characteristics (provided that the genes assort independently).

Now that you've had some experience working genetic crosses, explore Mendel's principles of heredity by setting up some of your own crosses in **Animation 3.1**.

The Dihybrid Testcross

Let's practice using the branch diagram by determining the types and proportions of phenotypes in a dihybrid testcross between the round and yellow F_1 pea plants (*Rr Yy*) obtained by Mendel in his dihybrid cross and the wrinkled and green pea plants (*rr yy*), as depicted in **Figure 3.12**. First, break the cross down into two single-locus crosses. The cross *Rr* × *rr* yields ½ round (*Rr*) progeny and ½ wrinkled (*rr*) progeny. The cross *Yy* × *yy* yields ½ yellow (*Yy*) progeny and ½ green (*yy*) progeny. Using the multiplication rule, we find the proportion of round and yellow progeny to be ½ (the probability of round) × ½ (the probability of yellow) = ¼. Four combinations of traits appear in the offspring in the following proportions: ¼ *Rr Yy*, round, yellow; ¼ *Rr yy*, round, green; ¼ *rr Yy*, wrinkled, yellow; and ¼ *rr yy*, wrinkled, green.



3.12 A branch diagram can be used to determine the phenotypes and expected proportions of offspring from a dihybrid testcross (*Rr Yy* × *rr yy*).

WORKED PROBLEM

The principles of segregation and independent assortment are important not only because they explain how heredity works but also because they provide the means for predicting the outcome of genetic crosses. This predictive power has made genetics a powerful tool in agriculture and other fields, and the ability to apply the principles of heredity is an important skill for all students of genetics. Practice with genetic problems is essential for mastering the basic principles of heredity; no amount of reading and memorization can substitute for the experience gained by deriving solutions to specific problems in genetics.

You may find genetics problems difficult if you are unsure of where to begin or how to organize a solution to the problem. In genetics, every problem is different, so no common series of steps can be applied to all genetics problems. Logic and common sense must be used to analyze a problem and arrive at a solution. Nevertheless, certain steps can facilitate the process, and solving the following problem will serve to illustrate those steps.

In mice, black coat color (*B*) is dominant to brown (*b*), and a solid pattern (*S*) is dominant to a white-spotted pattern (*s*). Color and spotting are controlled by genes that assort independently. A homozygous black, spotted mouse is crossed with a homozygous brown, solid mouse. All the F_1 mice are black and solid. A testcross is then carried out by mating the F_1 mice with brown, spotted mice.

a. Give the genotypes of the parents and the F_1 mice.

b. Give the genotypes and phenotypes, along with their expected ratios, of the progeny expected from the testcross.

SOLUTION STRATEGY

What information is required in your answer to the problem?

First, determine what question or questions the problem is asking. Is it asking for genotypes or genotypic ratios or phenotypic ratios? This problem asks you to provide the *genotypes* of the parents and the F_1, the *expected genotypes* and *phenotypes* of the progeny of the testcross, and their *expected proportions*.

What information is provided to solve the problem?

Next, determine what information is provided that will be necessary to solve the problem. This problem gives important information about the dominance relations of the traits involved and the genes that encode them:

- Black is dominant to brown.
- Solid is dominant to spotted.
- The genes for the two characteristics assort independently.
- Symbols for the different alleles: *B* for black, *b* for brown, *S* for solid, and *s* for spotted.

It is often helpful to write down the symbols at the beginning of the solution:

B—black *S—solid*
b—brown *s—spotted*

Next, write out the crosses given in the problem:

P	Homozygous black, spotted	×	Homozygous brown, solid
	↓		
F_1	Black, solid		
Testcross	Black, solid	×	Brown, spotted

For help with this problem, review:

If you need help solving the problem, review those sections of the chapter that cover the relevant information. For this problem, review Sections 3.2 and 3.3.

SOLUTION STEPS

STEP 1 Write down any genetic information that can be determined from the phenotypes alone.

From their phenotypes and the statement that they are homozygous, you know that the P-generation mice must be *BB ss* and *bb SS*. The F_1 mice are black and solid, both of which are dominant traits, so the F_1 mice must possess at least one black allele (*B*) and one solid allele (*S*). At this point, you may not be certain about the other alleles, so you can represent the genotype of the F_1 as *B_ S_*, where _ means that any allele is possible. The brown, spotted mice used in the testcross must be *bb ss* because both brown and spotted are recessive traits that will be expressed only if two recessive alleles are present. Record these genotypes on the crosses that you wrote out:

P	Homozygous black, spotted *BB ss*	×	Homozygous brown, solid *bb SS*
	↓		
F_1	Black, solid *B_ S_*		
Testcross	Black, solid *B_ S_*	×	Brown, spotted *bb ss*

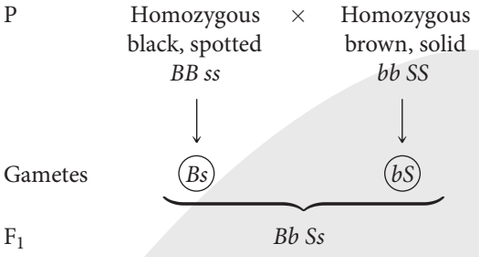
STEP 2 Break the problem down into smaller parts.

First, determine the genotype of the F_1. After this genotype has been determined, you can predict the results of the testcross and determine the genotypes and phenotypes of the progeny of the testcross. Second, because this cross includes two independently assorting loci, it can be conveniently broken down into two single-locus crosses: one for coat color and the other for spotting. Third, you can use a branch

diagram to determine the proportion of progeny of the testcross with different combinations of the two traits.

STEP 3 **Work the different parts of the problem.**

Start by determining the genotype of the F_1 progeny. Mendel's first law indicates that the two alleles at a locus separate, one going into each gamete. Thus, the gametes produced by the black, spotted parent contain *B s* and the gametes produced by the brown, solid parent contain *b S*, which combine to produce F_1 progeny with the genotype *Bb Ss*:



Use the F_1 genotype to work the testcross (*Bb Ss* × *bb ss*), breaking it into two single-locus crosses. First, consider the cross for coat color: *Bb* × *bb*. Any cross between a heterozygote and a homozygous recessive genotype produces a 1 : 1 phenotypic ratio of progeny (see Table 3.3):

Bb × *bb*

↓

½ *Bb* black
½ *bb* brown

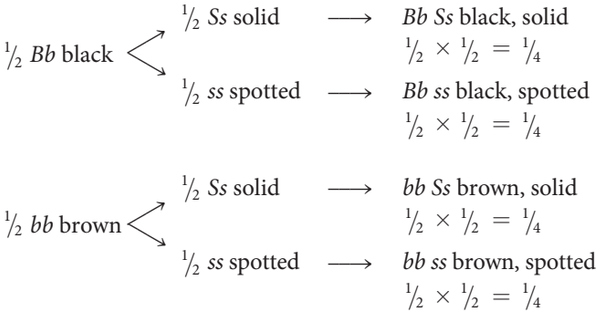
Next, consider the cross for spotting: *Ss* × *ss*. This cross is also between a heterozygote and a homozygous recessive genotype and produces ½ solid (*Ss*) and ½ spotted (*ss*) progeny (see Table 3.3):

Ss × *ss*

↓

½ *Ss* solid
½ *ss* spotted

Finally, determine the proportions of progeny with combinations of these characters by using a branch diagram.



STEP 4 **Check all work.**

As a last step, reread the problem, checking to see if your answers are consistent with the information provided. You have used the genotypes *BB ss* and *bb SS* in the P generation. Do these genotypes encode the phenotypes given in the problem? Are the F_1 progeny phenotypes consistent with the genotypes that you assigned? The answers are consistent with the information.

Now that we have stepped through a genetics problem together, try your hand at **Problem 31** at the end of the chapter.

3.4 Observed Ratios of Progeny May Deviate from Expected Ratios by Chance

When two individual organisms of known genotype are crossed, we expect certain ratios of genotypes and phenotypes among the progeny; these expected ratios are based on the Mendelian principles of segregation, independent assortment, and dominance. The ratios of genotypes and phenotypes *actually* observed among the progeny, however, may deviate from these expectations.

For example, in German cockroaches, brown body color (*Y*) is dominant to yellow body color (*y*). If we cross a brown, heterozygous cockroach (*Yy*) with a yellow cockroach (*yy*), we expect a 1 : 1 ratio of brown (*Yy*) and yellow (*yy*) progeny. Among 40 progeny, we therefore expect to see 20 brown and 20 yellow offspring. However, the observed numbers might deviate from these expected values; we might in fact see 22 brown and 18 yellow progeny.

Chance plays a critical role in genetic crosses, just as it does in flipping a coin. When you flip a coin, you expect a 1 : 1 ratio—½ heads and ½ tails. If you flipped a coin 1000 times, the proportion of heads and tails obtained would probably be very close to that expected 1 : 1 ratio. However, if you flipped the coin 10 times, the ratio of heads to tails might be quite different from 1 : 1. You could easily get 6 heads and 4 tails, or 3 heads and 7 tails, just by chance. You might even get 10 heads and 0 tails. The same thing happens in genetic crosses. We may expect 20 brown and 20 yellow cockroaches, but 22 brown and 18 yellow progeny *could* arise as a result of chance.

The Chi-Square (χ^2) Goodness-of-Fit Test

If you expected a 1 : 1 ratio of brown and yellow cockroaches but the cross produced 22 brown and 18 yellow

cockroaches, you probably wouldn't be too surprised, even though it wasn't a perfect 1 : 1 ratio. In this case, it seems reasonable to assume that chance produced the deviation between the expected and the observed results. But if you observed 25 brown and 15 yellow cockroaches, would you still assume that this result represents a 1 : 1 ratio? Something other than chance might have caused this deviation. Perhaps the inheritance of this characteristic is more complicated than was assumed, or perhaps some of the yellow progeny died before they were counted. Clearly, we need some means of evaluating how likely it is that chance is responsible for a deviation between the observed and the expected numbers.

To evaluate the role of chance in producing deviations between observed and expected values, a statistical test called the **chi-square goodness-of-fit test** is used. This test provides information about how well observed values fit expected values. Before we learn how to use this test, however, it is important to understand what it does and does not indicate about a genetic cross. The chi-square test cannot tell us whether a genetic cross has been correctly carried out, whether the results are correct, or whether we have chosen the correct genetic explanation for the results. What it does indicate is the *probability* that the difference between the observed and the expected values is due to chance. In other words, it indicates the likelihood that chance alone could produce the deviation between the expected and the observed values.

If we expected 20 brown and 20 yellow progeny from a genetic cross, the chi-square test gives us the probability that we might observe 25 brown and 15 yellow progeny simply owing to chance deviations from the expected 20 : 20 ratio. This hypothesis—that chance alone is responsible for a deviation between observed and expected values—is sometimes called the *null hypothesis*. Statistical tests such as the chi-square test cannot prove that the null hypothesis is correct, but they can help us decide whether we should reject it. When the probability calculated with the chi-square test is high, we assume that chance alone produced the deviation, and we do not reject the null hypothesis. When the probability is low, we assume that some factor other than chance—some significant factor—produced the deviation; for example, the mortality rate of the yellow cockroaches might be higher than that of the brown cockroaches. When the probability that chance produced the deviation is low, we reject the null hypothesis.

To use the chi-square goodness-of-fit test, we first determine the expected results. The chi-square test must always be applied to *numbers* of progeny, not to proportions or percentages. Let's consider a locus for coat color in domestic cats, for which black color (B) is dominant to gray (b). If we crossed two heterozygous black cats ($Bb \times Bb$), we would expect a 3 : 1 ratio of black and gray kittens. Imagine that a series of such crosses yields a total of 50 kittens—30 black and 20 gray. These numbers are our *observed* values. We can obtain the *expected* numbers by multiplying the expected proportions by the total number of observed progeny. In this case, the expected number of black kittens is $\frac{3}{4} \times 50 = 37.5$ and the expected number of gray kittens is $\frac{1}{4} \times 50 = 12.5$. Note that it is biologically impossible to have 37.5 or 12.5 kittens, because kittens only come in whole numbers. But the expected number is 37.5, so that is what we should use in the chi-square calculations.

The chi-square (χ^2) value is calculated by using the following formula:

$$\chi^2 = \sum \frac{(\text{observed} - \text{expected})^2}{\text{expected}}$$

where Σ means the sum. We calculate the sum of all the squared differences between observed and expected values divided by the expected values. To calculate the chi-square value for our black and gray kittens, we first subtract the number of *expected* black kittens from the number of *observed* black kittens ($30 - 37.5 = -7.5$) and square this value: $-7.5^2 = 56.25$. We then divide this result by the expected number of black kittens, $56.25/37.5 = 1.5$. We repeat the calculations on the number of expected gray kittens: $(20 - 12.5)^2/12.5 = 4.5$. To obtain the overall chi-square value, we sum the $(\text{observed} - \text{expected})^2/\text{expected}$ values: $1.5 + 4.5 = 6.0$.

The next step is to determine the probability associated with this calculated chi-square value, which is the probability that the deviation between the observed and the expected results is due to chance. This step requires us to compare our calculated chi-square value (6.0) with theoretical values in a chi-square table that have the same degrees of freedom. The degrees of freedom represent the number of ways in which the expected classes are free to vary. For a chi-square goodness-of-fit test, the number of degrees of freedom is equal to $n - 1$, in which n is the number of different expected phenotypes. Here, we lose one degree of freedom because the total number of expected progeny must equal the total number of observed progeny. In our example, there are two expected phenotypes (black and gray), so $n = 2$, and the degree of freedom equals $2 - 1 = 1$.

Now that we have our calculated chi-square value and have figured out the associated degrees of freedom, we are ready to obtain the probability from a chi-square table (**Table 3.5**). The degrees of freedom are given in the left-hand column of the table, and the probabilities are given at the top; within the body of the table are chi-square values associated with these probabilities. First, we find the row for the appropriate degrees of freedom; for our example with 1 degree of freedom, it is the first row of the table. Then, we find our calculated chi-square value (6.0) among the theoretical values in this row. The theoretical chi-square values increase, and the probabilities decrease, from left to right. Our chi-square value of 6.0 falls between

TABLE 3.5 Critical values of the χ^2 distribution

	P								
df	0.995	0.975	0.9	0.5	0.1	0.05*	0.025	0.01	0.005
1	0.000	0.000	0.016	0.455	2.706	3.841	5.024	6.635	7.879
2	0.010	0.051	0.211	1.386	4.605	5.991	7.378	9.210	10.597
3	0.072	0.216	0.584	2.366	6.251	7.815	9.348	11.345	12.838
4	0.207	0.484	1.064	3.357	7.779	9.488	11.143	13.277	14.860
5	0.412	0.831	1.610	4.351	9.236	11.070	12.832	15.086	16.750
6	0.676	1.237	2.204	5.348	10.645	12.592	14.449	16.812	18.548
7	0.989	1.690	2.833	6.346	12.017	14.067	16.013	18.475	20.278
8	1.344	2.180	3.490	7.344	13.362	15.507	17.535	20.090	21.955
9	1.735	2.700	4.168	8.343	14.684	16.919	19.023	21.666	23.589
10	2.156	3.247	4.865	9.342	15.987	18.307	20.483	23.209	25.188
11	2.603	3.816	5.578	10.341	17.275	19.675	21.920	24.725	26.757
12	3.074	4.404	6.304	11.340	18.549	21.026	23.337	26.217	28.300
13	3.565	5.009	7.042	12.340	19.812	22.362	24.736	27.688	29.819
14	4.075	5.629	7.790	13.339	21.064	23.685	26.119	29.141	31.319
15	4.601	6.262	8.547	14.339	22.307	24.996	27.488	30.578	32.801

P, probability; df, degrees of freedom.

*Most scientists assume that when $P < 0.05$, a significant difference exists between for parallel structure observed and expected values in a chi-square test.

the value of 5.024, associated with a probability of 0.025, and the value of 6.635, associated with a probability of 0.01.

Thus, the probability associated with our chi-square value is less than 0.025 and greater than 0.01. So there is less than a 2.5% probability that the deviation that we observed between the expected and the observed numbers of black and gray kittens could be due to chance.

Most scientists use the 0.05 probability level as their cutoff value: if the probability of chance being responsible for the deviation between observed and expected values is greater than or equal to 0.05, they accept that chance may be responsible for the deviation. When the probability is less than 0.05, scientists assume that chance is not responsible and that a significant difference from the expected values exists. The expression *significant difference* means that a factor other than chance is responsible for the deviation between the observed and the expected values. In regard to the kittens, perhaps one of the genotypes had a greater mortality rate before the progeny were counted, or perhaps other genetic factors skewed the observed ratios.

In choosing 0.05 as the cutoff value, scientists have agreed to assume that chance is responsible for deviations between observed and expected values unless there is strong evidence to the contrary. Bear in mind that even if we obtain a probability of, say, 0.01, there is still a 1% probability that the deviation between the observed and the expected values is due to nothing more than chance. Calculation of the chi-square value is illustrated in **Figure 3.13**. See **Animation 3.2** for an example of working a chi-square goodness-of-fit test. A

TRY PROBLEM 35

THINK-PAIR-SHARE Question 8

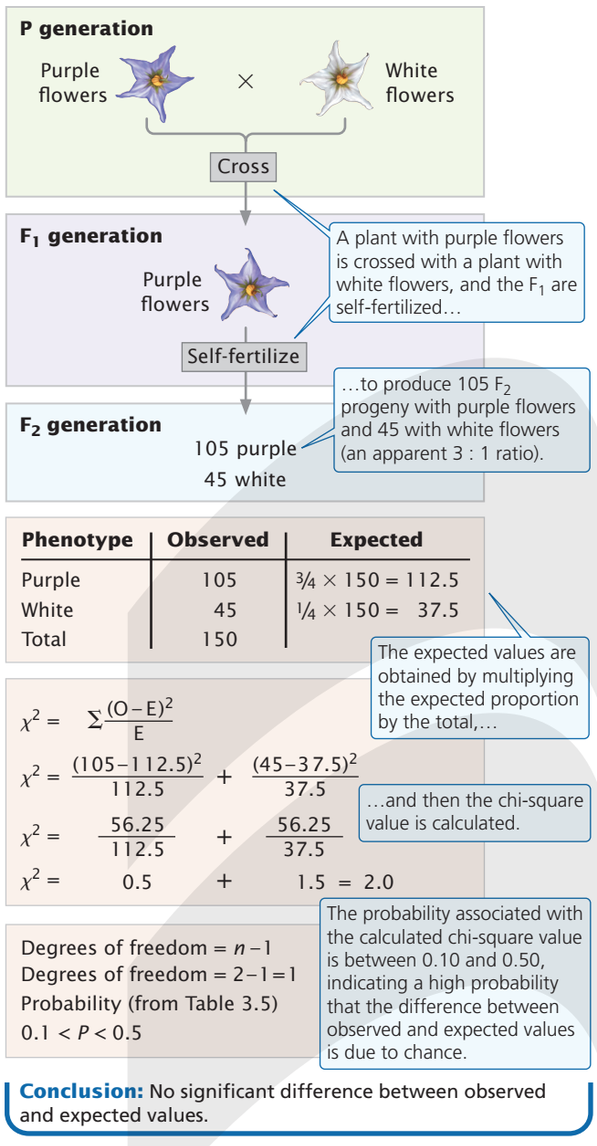
CONCEPTS

Differences between observed and expected numbers among the progeny of a cross can arise by chance alone. The chi-square goodness-of-fit test can be used to evaluate whether deviations between observed and expected values are likely to be due to chance or to some other, significant factor.

✓ CONCEPT CHECK 7

A chi-square test comparing observed and expected numbers of progeny is carried out, and the probability associated with the calculated chi-square value is 0.72. What does this probability represent?

a. Probability that the correct results were obtained
b. Probability of obtaining the observed numbers
c. Probability that the difference between observed and expected numbers is significant
d. Probability that the difference between observed and expected numbers could be due to chance



3.13 A chi-square goodness-of-fit test is used to determine the probability that the difference between observed and expected values is due to chance.

3.5 Geneticists Often Use Pedigrees to Study the Inheritance of Characteristics in Humans

The study of human genetic characteristics presents some major obstacles. First, controlled matings are not possible. With other organisms, geneticists can carry out specific crosses to test their hypotheses about inheritance. Unfortunately (for geneticists at least), matings between humans are more frequently determined by romance, family expectations, and—occasionally—chance encounters, than by the requirements of geneticists. Other obstacles are the long generation time and the generally small family size of our species. To overcome these obstacles, geneticists have developed techniques for studying human inheritance that are uniquely suited to human biology and culture.

One technique used by geneticists to study human inheritance is the analysis of pedigrees. A **pedigree** is a pictorial representation of a family history, essentially a family tree that outlines the inheritance of one or more characteristics. When a particular characteristic or disease is observed in a person, a geneticist often studies the family of this affected person by drawing a pedigree.

Symbols Used in Pedigrees

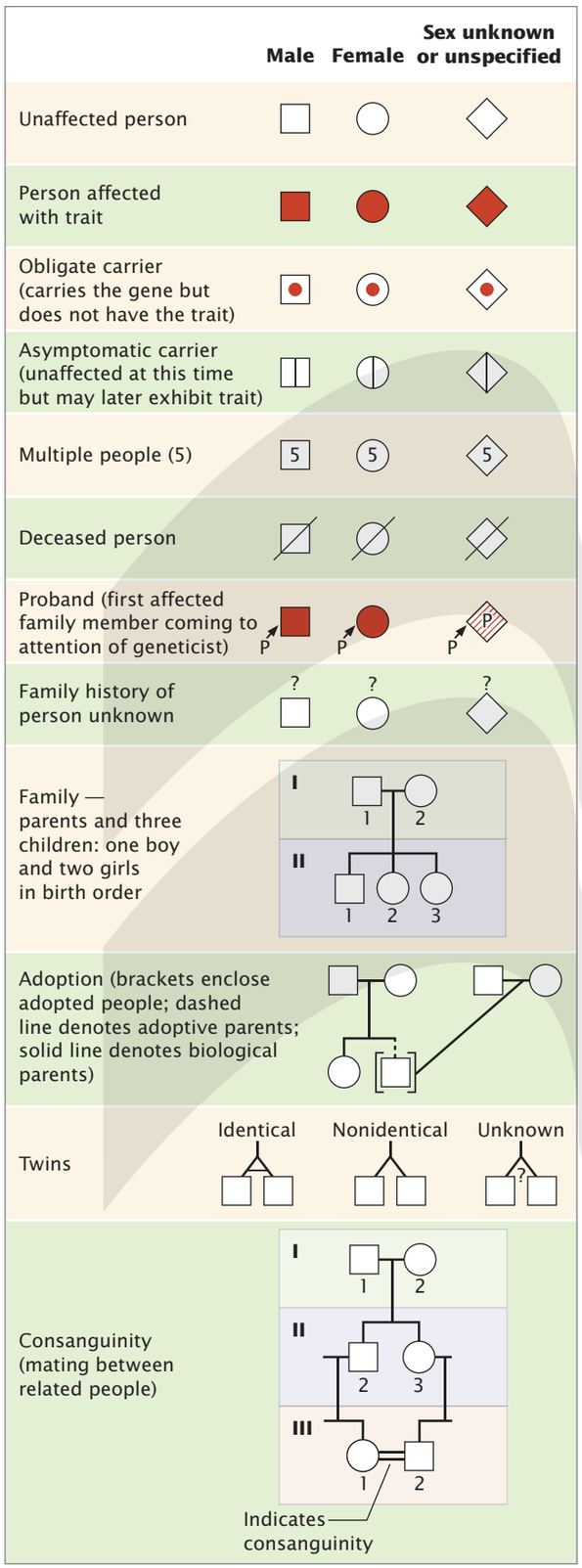
The symbols commonly used in pedigrees are summarized in **Figure 3.14**. Males in a pedigree are represented by squares, females by circles. A horizontal line drawn between two symbols representing a man and a woman indicates a mating; children are connected to their parents by vertical lines extending downward from the parents. The pedigree shown in **Figure 3.15a** illustrates a family with Waardenburg syndrome, an autosomal dominant type of deafness that may be accompanied by fair skin, a white forelock, and visual problems (**Figure 3.15b**). An autosomal trait is one that is encoded by a gene on an autosome (nonsex chromosome). In Chapter 4, we will consider the inheritance of sex-linked traits—those encoded by genes on the sex chromosomes.

Persons in a pedigree who exhibit the trait of interest are represented by filled circles and squares; in the pedigree of Figure 3.15a, the filled symbols represent members of the family who have Waardenburg syndrome. Unaffected members are represented by open circles and squares. The person from whom the pedigree is initiated is called the **proband** and is usually designated by the letter P and an arrow (IV-2 in Figure 3.15a).

Let's look closely at Figure 3.15 and consider some additional features of a pedigree. Each generation in a pedigree is identified by a roman numeral; within each generation, family members are assigned arabic numerals, and children in each family are listed in birth order from left to right. Person II-4, a man with Waardenburg syndrome, mated with II-5, an unaffected woman, and they produced five children. The oldest of their children is III-8, a male with Waardenburg syndrome, and the youngest is III-14, an unaffected female.

Analysis of Pedigrees

The limited number of offspring in most human families means that clear Mendelian ratios are usually impossible to discern in a single pedigree. Pedigree analysis requires a



3.14 Standard symbols are used in pedigrees.

certain amount of genetic sleuthing, based on recognizing patterns associated with different modes of inheritance.

Recessive Traits Autosomal recessive traits normally appear with equal frequency in both sexes and appear only when a person inherits two alleles for the trait, one from each parent. If the trait is uncommon, most parents of affected offspring are heterozygous and unaffected; consequently, the trait seems to skip generations (**Figure 3.16**). Frequently, a recessive allele may be passed on for a number of generations without the trait appearing in a pedigree. When both parents are heterozygous, approximately one-fourth of the offspring are expected to express the trait, but this ratio will not be obvious unless the family is large. In the rare event that both parents are affected by an autosomal recessive trait, all the offspring will be affected.

When a recessive trait is rare, most people outside affected families are homozygous for the normal allele. Thus, when an affected person mates with someone outside the family ($aa \times AA$), usually none of the children display the trait, although all will be carriers (i.e., heterozygous). A recessive trait is more likely to appear in a pedigree when two people within the same family mate, because there is a greater chance of both parents carrying the same recessive allele. Mating between closely related people is called **consanguinity**.

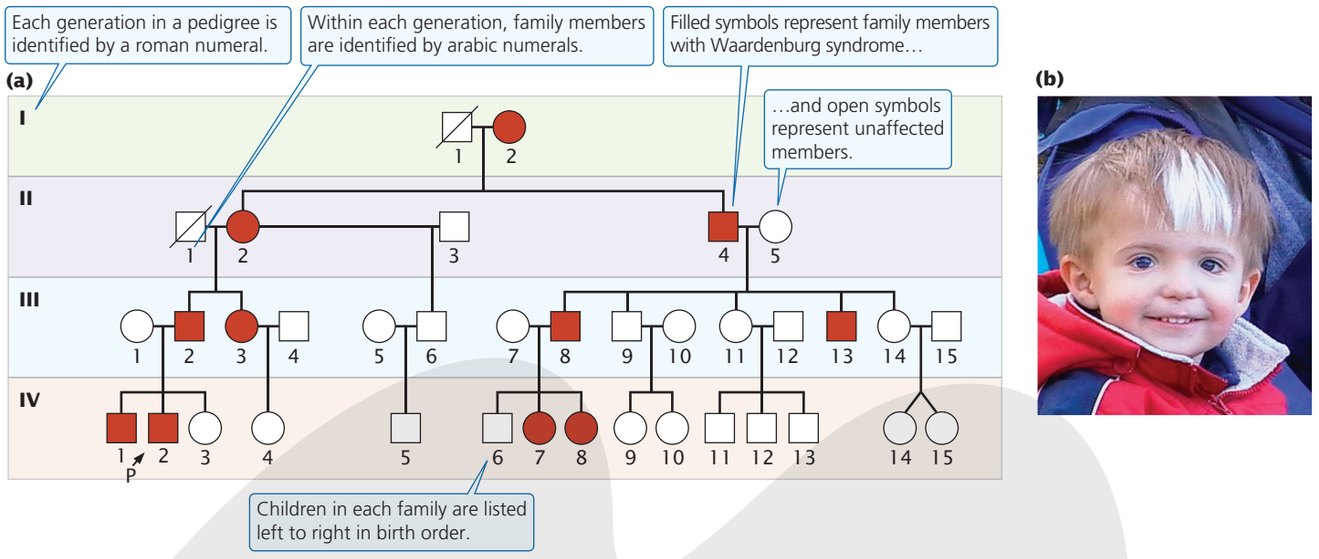
CONCEPTS

Autosomal recessive traits appear with equal frequency in males and females. Affected children are commonly born to unaffected parents who are heterozygous carriers of the gene for the trait, and the trait tends to skip generations. Recessive traits appear more frequently among the offspring of consanguineous matings. Autosomal dominant traits also appear in both sexes with equal frequency. An affected person has an affected parent, and the trait does not skip generations. Unaffected persons do not transmit the trait.

✓ CONCEPT CHECK 8

Recessive traits often appear in pedigrees in which there have been consanguineous matings because these traits

a. tend to skip generations.
b. appear only when both parents carry a copy of the gene for the trait, which is more likely when the parents are related.
c. usually arise in children born to parents who are unaffected.
d. appear equally in males and females.

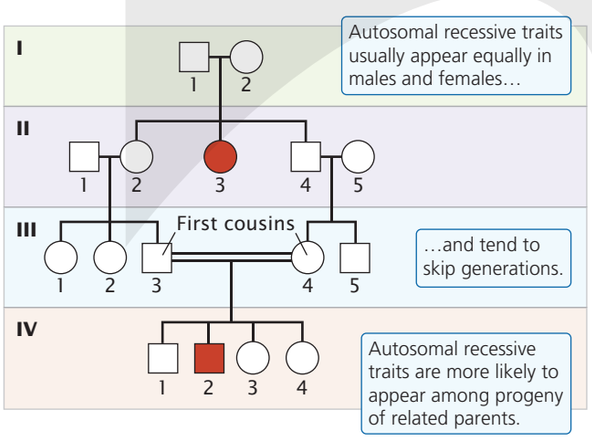


3.15 Waardenburg syndrome is (a) inherited as an autosomal dominant trait and (b) characterized by deafness, fair skin, visual problems, and a white forelock.
The proband (P) is the person from whom this pedigree is initiated. [Photograph courtesy of Guy Rowland.]

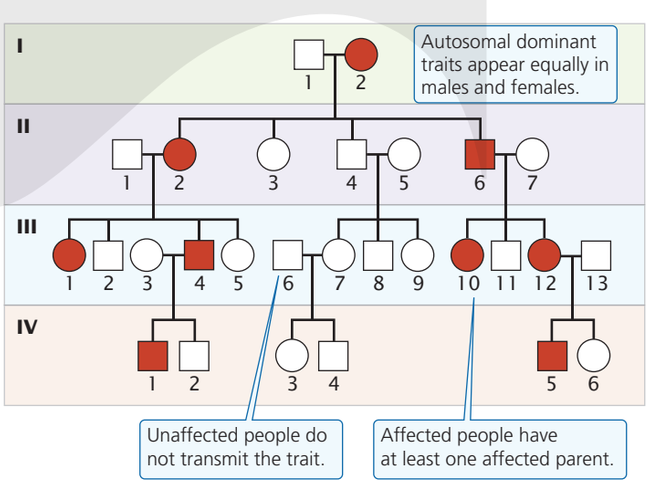
In the pedigree shown in Figure 3.16, individuals III-3 and III-4 are first cousins, and both are heterozygous for the recessive allele; when two heterozygotes mate, one-fourth of their children are expected to have the recessive trait.

Dominant Traits Autosomal dominant traits appear in both sexes with equal frequency, and both sexes are capable of transmitting these traits to their offspring. Every person with a dominant trait must have inherited the allele from at least one parent; autosomal dominant traits therefore do not skip generations (**Figure 3.17**).

Sex-linked traits also have a distinctive pattern of inheritance, which we will consider in Chapter 4. See the analysis of a pedigree in **Animation 3.3**. TRY PROBLEM 38 A



3.16 Recessive traits normally appear with equal frequency in both sexes and often skip generations.
The double line between III-3 and III-4 represents consanguinity (mating between related persons).



3.17 Dominant traits normally appear with equal frequency in both sexes and do not skip generations.

CHAPTER SUMMARY

Section 3.1 Learning Objective: Be able to explain how Gregor Mendel discovered the principles of heredity.

- Gregor Mendel discovered the principles of heredity. His success can be attributed to his choice of the pea plant as an experimental organism, the use of characteristics with a few easily distinguishable phenotypes, his experimental approach, the use of mathematics to interpret his results, and careful attention to detail.
- Genes are inherited factors that determine a characteristic. Alternative forms of a gene are called alleles. The alleles are located at a specific place, called a locus, on a chromosome, and the set of alleles that an individual organism possesses is its genotype. A phenotype is the manifestation or appearance of a characteristic and may refer to a physical, physiological, biochemical, or behavioral characteristic. Only the genotype—not the phenotype—is inherited.

Section 3.2 Learning Objective: Be able to use principles of heredity to predict progeny produced in a simple genetic cross.

- The principle of segregation states that a diploid individual organism possesses two alleles encoding a trait and that these two alleles separate in equal proportions when gametes are formed.
- The concept of dominance indicates that when two different alleles are present in a heterozygote, only the trait encoded by one of them, the dominant allele, is observed in the phenotype. The other allele is said to be recessive.
- The two alleles of a genotype are located on homologous chromosomes. The separation of homologous chromosomes in anaphase I of meiosis brings about the segregation of alleles.
- Probability is the likelihood that a particular event will occur. The multiplication rule states that the probability of two or more independent events occurring together is calculated by multiplying the probabilities of the independent events. The addition rule states that the probability of any of two or more mutually exclusive events occurring is calculated by adding the probabilities of the events.
- A testcross, which reveals the genotype (homozygote or heterozygote) of an individual organism that has a dominant trait, consists of crossing that individual with one that has the homozygous recessive genotype.

Section 3.3 Learning Objective: Be able to predict the progeny of crosses involving two or more loci.

- The principle of independent assortment states that genes encoding different characteristics separate independently when gametes are formed. Independent assortment is based on the random separation of homologous pairs of chromosomes in anaphase I of meiosis; it takes place when genes encoding different characteristics are located on different pairs of chromosomes.

Section 3.4 Learning Objective: Be able to use the chi-square goodness-of-fit test to determine the probability that observed numbers of progeny differ from expected numbers.

- Observed ratios of progeny from a genetic cross may deviate from the expected ratios owing to chance. The chi-square goodness-of-fit test can be used to determine the probability that a difference between observed and expected numbers could be due to chance.

Section 3.5 Learning Objective: Be able to use pedigrees to analyze the genetic basis of traits in humans.

- Special symbols and formats are used to construct pedigrees.
- Autosomal recessive traits typically appear with equal frequency in both sexes and tend to skip generations. When both parents are heterozygous for a particular autosomal recessive trait, approximately one-fourth of their offspring will have the trait. Recessive traits are more likely to appear in families with consanguinity (mating between closely related people).
- Autosomal dominant traits usually appear equally in both sexes and do not skip generations. When one parent is affected and heterozygous for an autosomal dominant trait, approximately half of the offspring will have the trait. Unaffected people do not normally transmit an autosomal dominant trait to their offspring.

IMPORTANT TERMS

gene (p. 49)
allele (p. 49)
locus (loci) (p. 49)
genotype (p. 49)
homozygous (p. 49)
heterozygous (p. 49)
phenotype (p. 49)
monohybrid cross (p. 50)
P (parental) generation (p. 50)
F_1 (first filial) generation (p. 50)
reciprocal cross (p. 50)
F_2 (second filial) generation (p. 50)
dominant (p. 51)
recessive (p. 51)
principle of segregation (Mendel's first law) (p. 52)
concept of dominance (p. 52)

chromosome theory of heredity (p. 53)
backcross (p. 54)
Punnett square (p. 55)
probability (p. 55)
multiplication rule (p. 56)
addition rule (p. 56)
conditional probability (p. 57)
testcross (p. 57)
wild type (p. 58)
dihybrid cross (p. 59)
principle of independent assortment (Mendel's second law) (p. 59)
chi-square goodness-of-fit test (p. 65)
pedigree (p. 67)
proband (p. 67)
consanguinity (p. 68)

ANSWERS TO CONCEPT CHECKS

1. b

2. A locus is a place on a chromosome where genetic information encoding a characteristic is located. An allele is a version of a gene that encodes a specific trait. A genotype is the set of alleles possessed by an individual organism, and a phenotype is the manifestation or appearance of a characteristic.

3. The traits encoded by both alleles appeared in the F_2 progeny.

4. d

5. a

6. Both the principle of segregation and the principle of independent assortment refer to the separation of alleles in anaphase I of meiosis. The principle of segregation says that these alleles separate, and the principle of independent assortment says that they separate independently of alleles at other loci.

7. d

8. b

WORKED PROBLEMS

Problem 1

In corn, purple kernels are dominant to yellow kernels, and full kernels are dominant to shrunken kernels. A corn plant that has purple and full kernels is crossed with a plant that has yellow and shrunken kernels, and the following progeny are obtained:

purple, full	112
purple, shrunken	103
yellow, full	91
yellow, shrunken	94

What are the most likely genotypes of the parents and progeny? Test your genetic hypothesis with a chi-square test.

Solution Strategy

What information is required in your answer to the problem?

a. The genotypes of parents and progeny.

b. A chi-square test comparing the observed and expected results and the interpretation of the chi-square test.

What information is provided to solve the problem?

- Purple kernels are dominant to yellow kernels, and full kernels are dominant to shrunken kernels.
- The phenotypes of the parents.
- The phenotypes and numbers of progeny of the cross.

For help with this problem, review:

Sections 3.3 and 3.4.

Solution Steps

The best way to begin this problem is by breaking the cross down into simple crosses for a single characteristic (kernel color or kernel shape):

P	purple × yellow	full × shrunken
F_1	112 + 103 = 215 purple	112 + 91 = 203 full
	91 + 94 = 185 yellow	103 + 94 = 197 shrunken

In this cross, purple × yellow produces approximately ½ purple and ½ yellow (a 1 : 1 ratio). A 1 : 1 ratio is usually the result of a cross between a heterozygote and a homozygote. Because purple is dominant, the purple parent must be heterozygous (*Pp*) and the yellow parent must be homozygous (*pp*). The purple progeny produced by this cross will be heterozygous (*Pp*), and the yellow progeny must be homozygous (*pp*).

Hint: A good strategy in a cross involving multiple characteristics is to analyze the results for each characteristic separately.

Recall: The multiplication rule states that the probability of two or more independent events occurring together is calculated by multiplying their independent probabilities.

Now let's examine the other character. Full × shrunken produces ½ full and ½ shrunken, or a 1 : 1 ratio, so these progeny phenotypes are also produced by a cross between a heterozygote (*Ff*) and a homozygote (*ff*); the full-kernel progeny will be heterozygous (*Ff*), and the shrunken-kernel progeny will be homozygous (*ff*).

Now combine the two crosses and use the multiplication rule to obtain the overall genotypes and the proportions of each genotype:

P Purple, full × Yellow, shrunken
Pp Ff *pp ff*

F_1 *Pp Ff* = ½ purple × ½ full = ¼ purple, full
Pp ff = ½ purple × ½ shrunken = ¼ purple, shrunken
pp Ff = ½ yellow × ½ full = ¼ yellow, full
pp ff = ½ yellow × ½ shrunken = ¼ yellow, shrunken

Our genetic calculations predict that from this cross, we should see ¼ purple, full-kernel progeny; ¼ purple, shrunken-kernel progeny; ¼ yellow, full-kernel progeny; and ¼ yellow, shrunken-kernel progeny. A total of 400 progeny were produced, so $\frac{1}{4} \times 400 = 100$ of each phenotype are expected. Therefore, the observed numbers do not fit the expected numbers exactly.

Could the difference between what we observe and what we expected be due to chance? If the probability is high that chance alone is responsible for the difference between observed and expected values, we will assume that the progeny have been produced in the 1 : 1 : 1 : 1 ratio predicted for the cross. If the probability is low that the difference between observed and expected values is due to chance, we will assume that the progeny really are not in the predicted ratio and that some other, *significant* factor must be responsible for the deviation from our expectations.

The observed and expected numbers are

Phenotype	Observed	Expected
purple, full	112	$\frac{1}{4} \times 400 = 100$
purple, shrunken	103	$\frac{1}{4} \times 400 = 100$
yellow, full	91	$\frac{1}{4} \times 400 = 100$
yellow, shrunken	94	$\frac{1}{4} \times 400 = 100$

To determine the probability that the difference between observed and expected numbers is due to chance, we calculate a chi-square value using the formula

Hint: See Figure 3.13 for help on how to carry out a chi-square test.

$$\chi^2 = \Sigma\,[(\text{observed} - \text{expected})^2/\text{expected}]:$$

$$\chi^2 = \frac{(112-100)^2}{100} + \frac{(103-100)^2}{100} + \frac{(91-100)^2}{100} + \frac{(94-100)^2}{100}$$

$$= \frac{12^2}{100} + \frac{3^2}{100} + \frac{9^2}{100} + \frac{6^2}{100}$$

$$= \frac{144}{100} + \frac{9}{100} + \frac{81}{100} + \frac{36}{100}$$

$$= 1.44 + 0.09 + 0.81 + 0.36 = 2.70$$

The next step is to determine the probability associated with this calculated chi-square value, which is the probability that the deviation between the observed and the expected results is due to chance. This step requires us to compare our calculated chi-square value (2.70) with theoretical values in a chi-square table that have the same degrees of freedom. The degrees of freedom for a chi-square goodness-of-fit test are $n - 1$, where n equals the number of expected phenotypic classes. In this case, there are four expected phenotypic classes, so the degrees of freedom equal $4 - 1 = 3$. We must now look up the chi-square value in a chi-square table (see Table 3.5). We select the row corresponding to 3 degrees of freedom and look along this row to find our calculated chi-square value. The calculated chi-square value of 2.7 lies between 2.366 (a probability of 0.5) and 6.251 (a probability of 0.1). Therefore, the probability (P) associated with the calculated chi-square value is $0.5 > P > 0.1$. This P is the probability that the difference between what we observed and what we expected is due to chance, which in this case is relatively high, so chance is probably responsible for the deviation. We can conclude that the progeny *do* appear in the 1 : 1 : 1 : 1 ratio predicted by our genetic explanation.

Problem 2

Joanna has "short fingers" (brachydactyly). She has two older brothers who are identical twins; both have short fingers. Joanna's two younger sisters have normal fingers. Joanna's mother has normal fingers, and her father has short fingers. Joanna's paternal grandmother (her father's mother) has short fingers; her paternal grandfather (her father's father), who is now deceased, had normal fingers. Both of Joanna's maternal grandparents (her mother's parents) have normal fingers. Joanna marries Tom, who has normal fingers; they adopt a son named Bill, who has normal fingers. Bill's biological parents both have normal fingers. After adopting Bill, Joanna and

Tom produce two children: an older daughter with short fingers and a younger son with normal fingers.

a. Using standard symbols and labels, draw a pedigree illustrating the inheritance of short fingers in Joanna's family.

b. What is the most likely mode of inheritance for short fingers in this family?

c. If Joanna and Tom have another biological child, what is the probability (based on your answer to part *b*) that this child will have short fingers?

Solution Strategy

What information is required in your answer to the problem?

a. A pedigree to represent the family, drawn with correct symbols and labeling.

b. The most likely mode of inheritance for short fingers.

c. The probability that Joanna and Tom's next child will have short fingers.

What information is provided to solve the problem?

The phenotypes of Joanna and Tom and their family members.

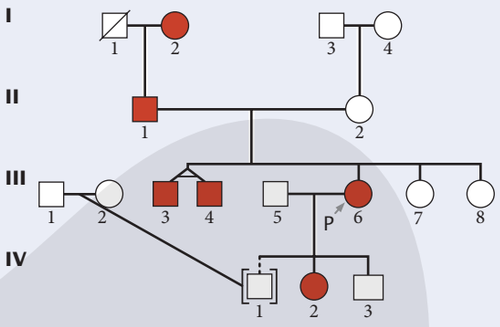
For help with this problem, review:

The information on pedigrees in Section 3.5.

Solution Steps

Hint: See Figure 3.14 for a review of symbols used in a pedigree.

a. In the pedigree for the family, use filled circles (females) and filled squares (males) to represent family members with the trait of interest (short fingers). Connect Joanna's identical twin brothers to the line above by drawing diagonal lines that have a horizontal line between them. Enclose Bill, the adopted child of Joanna and Tom, in brackets; connect him to his biological parents by drawing a diagonal line and to his adoptive parents by a dashed line.



b. The most likely mode of inheritance for short fingers in this family is autosomal dominant. The trait appears equally in males and females and does not skip generations. When one parent has the trait, it appears in approximately half of that parent's sons and daughters, although the number of children in the families is small.

Hint: See Analysis of Pedigrees for a review of characteristics of dominant and recessive traits in pedigrees.

c. If having short fingers is an autosomal dominant trait, Tom must be homozygous (*bb*) because he has normal fingers. Joanna must be heterozygous (*Bb*) because she and Tom have produced both short-fingered and normal-fingered offspring. In a cross between a heterozygote and homozygote, half the progeny are expected to be heterozygous and the other half homozygous ($Bb \times bb \rightarrow \frac{1}{2}Bb, \frac{1}{2}bb$), so the probability that Joanna and Tom's next biological child will have short fingers is $\frac{1}{2}$.

COMPREHENSION QUESTIONS

Section 3.1

1. Why was Mendel's approach to the study of heredity so successful?

2. What is the difference between genotype and phenotype?

Section 3.2

3. What is the principle of segregation? Why is it important?

4. How are Mendel's principles different from the concept of blending inheritance discussed in Chapter 1?

5. What is the concept of dominance?

6. What are the addition and multiplication rules of probability, and when should they be used?

7. Give the genotypic ratios that may appear among the progeny of simple crosses and the genotypes of the parents that may give rise to each ratio.

8. What is the chromosome theory of heredity? Why was it important?

Section 3.3

9. What is the principle of independent assortment? How is it related to the principle of segregation?

10. In which stages of mitosis and meiosis are the principles of segregation and independent assortment at work?

Section 3.4

11. How is the chi-square goodness-of-fit test used to analyze genetic crosses? What does the probability associated with a chi-square value indicate about the results of a cross?

Section 3.5

12. What features are exhibited by a pedigree of a recessive trait? What features are exhibited if the trait is dominant?

For more questions that test your comprehension of the key chapter concepts, go to LearningCurve macmillan learning for this chapter.

APPLICATION QUESTIONS AND PROBLEMS

Introduction

13. If blond hair in the Solomon Islanders had originated from early European explorers, what would you predict the researchers would have found when they conducted their genetic study of the islanders?

Section 3.1

***14.** What characteristics of an organism would make it suitable for studies of the principles of inheritance? Name several organisms that have these characteristics.

Section 3.2

15. In cucumbers, orange fruit color (*R*) is dominant to cream fruit color (*r*). A cucumber plant homozygous for orange fruit is crossed with a plant homozygous for cream fruit. The F_1 are intercrossed to produce the F_2.

a. Give the genotypes and phenotypes of the parents, the F_1, and the F_2.

b. Give the genotypes and phenotypes of the offspring of a backcross between the F_1 and the orange-fruited parent.

c. Give the genotypes and phenotypes of a backcross between the F_1 and the cream-fruited parent.

16. **Figure 1.1** (p. 2) shows three girls, one of whom has albinism. Could the three girls shown in the photograph be sisters? Why or why not?

17. DATA ANALYSIS J. W. McKay crossed a stock melon plant that produced tan seeds with a plant that produced red seeds and obtained the following results (J. W. McKay. 1936. *Journal of Heredity* 27:110–112).

Cross	F_1	F_2
tan ♀ × red ♂	13 tan seeds	93 tan seeds, 24 red seeds

a. Explain the inheritance of tan and red seeds in this plant.

b. Assign symbols for the alleles in this cross, and give genotypes for all the individual plants.

18. DATA ANALYSIS White (*w*) coat color in guinea pigs is recessive to black (*W*). In 1909, W. E. Castle and J. C. Phillips transplanted an ovary from a black guinea pig into a white female whose ovaries had been removed. They then mated this white female with a white male. All the offspring from the mating were black (W. E. Castle and J. C. Phillips. 1909. *Science* 30:312–313).


[Left: © Wegner/ARCO/Nature Picture Library. Right: © Nigel Cattlin/Alamy.]

a. Explain the results of this cross.

b. Give the genotype of the offspring of this cross.

c. What, if anything, does this experiment indicate about the validity of the pangenesis and the germ-plasm theories discussed in Chapter 1?

***19.** In cats, blood type A results from an allele (I^A) that is dominant to an allele (i^B) that produces blood type B. There is no O blood type. The blood types of male and female cats that were mated and the blood types of their kittens follow. Give the most likely genotypes for the parents of each litter.

	Male parent	Female parent	Kittens
a.	A	B	4 with type A, 3 with type B
b.	B	B	6 with type B
c.	B	A	8 with type A
d.	A	A	7 with type A, 2 with type B
e.	A	A	10 with type A
f.	A	B	4 with type A, 1 with type B

20. **Figure 3.7** shows the results of a cross between a tall pea plant and a short pea plant.

a. What phenotypes and proportions will be produced if a tall F_1 plant is backcrossed to the short parent?

b. What phenotypes and proportions will be produced if a tall F_1 plant is backcrossed to the tall parent?

21. Joe has a white cat named Sam. When Joe crosses Sam with a black cat, he obtains ½ white kittens and ½ black

kittens. When the black kittens are interbred, all the kittens that they produce are black. On the basis of these results, would you conclude that white or black coat color in cats is a recessive trait? Explain your reasoning.

***22.** Alkaptonuria is a metabolic disorder in which affected people produce black urine. Alkaptonuria results from an allele (*a*) that is recessive to the allele for normal metabolism (*A*). Sally has normal metabolism, but her brother has alkaptonuria. Sally's father has alkaptonuria, and her mother has normal metabolism.

a. Give the genotypes of Sally, her mother, her father, and her brother.

b. If Sally's parents have another child, what is the probability that this child will have alkaptonuria?

c. If Sally marries a man with alkaptonuria, what is the probability that their first child will have alkaptonuria?

23. Hairlessness in American rat terriers is recessive to the presence of hair. Suppose that you have a rat terrier with hair. How can you determine whether this dog is homozygous or heterozygous for the hairy trait?

***24.** What is the probability of rolling one six-sided die and obtaining the following numbers?

a. 2

b. 1 or 2

c. An even number

d. Any number but a 6

***25.** What is the probability of rolling two six-sided dice and obtaining the following numbers?

a. 2 and 3

b. 6 and 6

c. At least one 6

d. Two of the same number (two 1s, two 2s, two 3s, etc.)

e. An even number on both dice

f. An even number on at least one die

26. Phenylketonuria (PKU) is a disease that results from a recessive gene. Suppose that two unaffected parents produce a child with PKU.

a. What is the probability that a sperm from the father will contain the PKU allele?

b. What is the probability that an egg from the mother will contain the PKU allele?

c. What is the probability that their next child will have PKU?

d. What is the probability that their next child will be heterozygous for the PKU gene?

***27.** In German cockroaches, curved wings (*cv*) are recessive to normal wings (cv^+). A homozygous cockroach having normal wings is crossed with a homozygous cockroach having curved wings. The F_1 are intercrossed to produce the F_2. Assume that the pair of chromosomes containing the locus for wing shape is metacentric. Draw this pair of chromosomes as it would appear in the parents, the F_1, and each class of F_2 progeny at metaphase I of meiosis. Assume that no crossing over takes place. At each stage, label a location for the alleles for wing shape (*cv* and cv^+) on the chromosomes. See Chapter 2 for a review of meiosis.

***28.** In guinea pigs, the allele for black fur (*B*) is dominant to the allele for brown fur (*b*). A black guinea pig is crossed with a brown guinea pig, producing five F_1 black guinea pigs and six F_1 brown guinea pigs.

a. How many copies of the black allele (*B*) will be present in each cell of an F_1 black guinea pig at the following stages: G_1, G_2, metaphase of mitosis, metaphase I of meiosis, metaphase II of meiosis, and after the second cytokinesis following meiosis? Assume that no crossing over takes place. See Chapter 2 for discussion of mitosis and meiosis.

b. How many copies of the brown allele (*b*) will be present in each cell of an F_1 brown guinea pig at the same stages as those listed in part *a*? Assume that no crossing over takes place.

Section 3.3

29. In watermelons, bitter fruit (*B*) is dominant to sweet fruit (*b*), and yellow spots (*S*) are dominant to no spots (*s*). The genes for these two characteristics assort independently. A homozygous plant that has bitter fruit and yellow spots is crossed with a homozygous plant that has sweet fruit and no spots. The F_1 are intercrossed to produce the F_2.

a. What are the phenotypic ratios in the F_2?

b. If an F_1 plant is backcrossed with the bitter, yellow-spotted parent, what phenotypes and proportions are expected in the offspring?

c. If an F_1 plant is backcrossed with the sweet, unspotted parent, what phenotypes and proportions are expected in the offspring?

30. **Figure 3.9** shows the results of a dihybrid cross involving seed shape and seed color.

a. What proportion of the round and yellow F_2 progeny from this cross is homozygous at both loci?

b. What proportion of the round and yellow F_2 progeny from this cross is homozygous at least at one locus?

***31.** In cats, curled ears result from an allele (*Cu*) that is dominant to an allele (*cu*) for normal ears. Black color results from an independently assorting allele (*G*) that is dominant to an allele for gray (*g*). A gray cat homozygous for

[Jean-Michel Labat/Science Source.]

curled ears is mated with a homozygous black cat with normal ears. All the F_1 cats are black and have curled ears.

a. If two of the F_1 cats mate, what phenotypes and proportions are expected in the F_2?

b. An F_1 cat mates with a stray cat that is gray and possesses normal ears. What phenotypes and proportions of progeny are expected from this cross?

***32.** The following two genotypes are crossed: *Aa Bb Cc dd Ee* × *Aa bb Cc Dd Ee*. What will be the proportion of the following genotypes among the progeny of this cross?

a. *Aa Bb Cc Dd Ee* **b.** *Aa bb Cc dd ee*

c. *aa bb cc dd ee* **d.** *AA BB CC DD EE*

33. In cucumbers, dull fruit (*D*) is dominant to glossy fruit (*d*), orange fruit (*R*) is dominant to cream fruit (*r*), and bitter cotyledons (*B*) are dominant to non-bitter cotyledons (*b*). The three characters are encoded by genes located on different pairs of chromosomes. A plant homozygous for dull, orange fruit and bitter cotyledons is crossed with a plant that has glossy, cream fruit and non-bitter cotyledons. The F_1 are intercrossed to produce the F_2.

a. Give the phenotypes and their expected proportions in the F_2.

b. An F_1 plant is crossed with a plant that has glossy, cream fruit and non-bitter cotyledons. Give the phenotypes and expected proportions among the progeny of this cross.

***34.** Alleles *A* and *a* are located on a pair of metacentric chromosomes. Alleles *B* and *b* are located on a pair of acrocentric chromosomes (see **Figure 2.7** in Chapter 2). A cross is made between individuals having the following genotypes: *Aa Bb* × *aa bb*.

a. Draw the chromosomes as they would appear in each type of gamete produced by these individuals.

b. For each type of progeny resulting from this cross, draw the chromosomes as they would appear in a cell at G_1, G_2, and metaphase of mitosis.

Section 3.4

***35.** J. A. Moore investigated the inheritance of spotting patterns in leopard frogs (J. A. Moore. 1943. *Journal of Heredity* 34:3–7). The pipiens phenotype had the normal spots that give leopard frogs their name. In contrast, the burnsi phenotype lacked spots on its back. Moore carried out the following crosses, producing the progeny indicated.

Parent phenotypes	Progeny phenotypes
burnsi × burnsi	39 burnsi, 6 pipiens
burnsi × pipiens	23 burnsi, 33 pipiens
burnsi × pipiens	196 burnsi, 210 pipiens

a. On the basis of these results, is the burnsi phenotype most likely inherited as a dominant trait or as a recessive trait?

b. Give the most likely genotypes of the parent in each cross (use *B* for the burnsi allele and B^+ for the pipiens allele).

c. Use a chi-square test to evaluate the fit of the observed numbers of progeny to the number expected on the basis of your proposed genotypes.

***36.** In the California poppy, an allele for yellow flowers (*C*) is dominant to an allele for white flowers (*c*). At an independently assorting locus, an allele for entire petals (*F*) is dominant to an allele for fringed petals (*f*). A plant that is homozygous for yellow and entire petals is crossed with a plant that has white and fringed petals. A resulting F_1 plant is then crossed with a plant that has white and fringed petals, and the following progeny are produced: 54 yellow and entire; 58 yellow and fringed; 53 white and entire; 10 white and fringed.

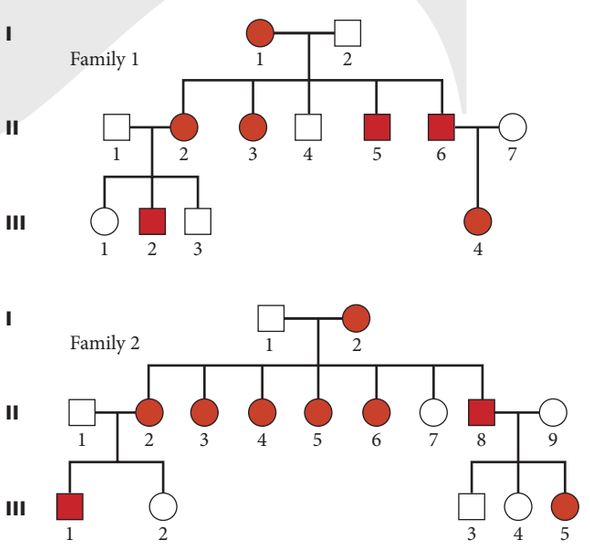
a. Use a chi-square test to compare the observed numbers of progeny having each phenotype with those expected for the cross.

b. What conclusion can you draw from the results of the chi-square test?

c. Suggest an explanation for the results.

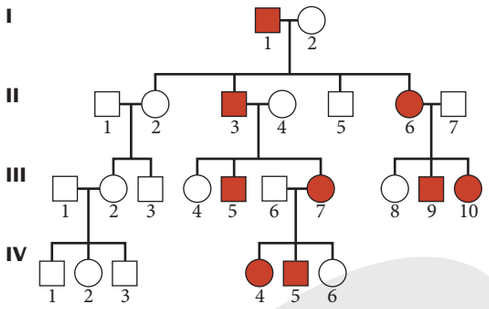
Section 3.5

37. Many studies have suggested a strong genetic predisposition to migraine headaches, but the mode of inheritance is not clear. L. Russo and colleagues examined migraine headaches in several families, two of which are shown below (L. Russo et al. 2005. *American Journal of Human Genetics* 76:327–333). What is the most likely mode of inheritance for migraine headaches in these families? Explain your reasoning.

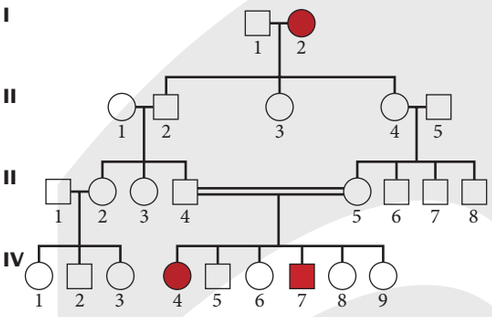


*38. For each of the following pedigrees, give the most likely mode of inheritance, assuming that the trait is rare. Carefully explain your reasoning.

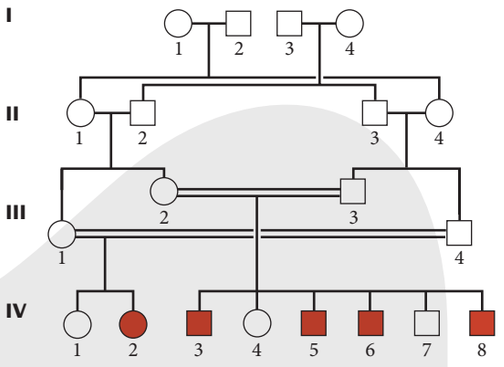
a.



b.



39. DATA ANALYSIS Ectrodactyly is a rare condition in which the fingers are absent and the hand is split. This condition is usually inherited as an autosomal dominant trait. Ademar Freire-Maia reported the appearance of ectrodactyly in a family in São Paulo, Brazil, whose pedigree is shown here. Is this pedigree consistent with autosomal dominant inheritance? If not, what mode of inheritance is most likely? Explain your reasoning.



[Data from A. Freire-Maia, *Journal of Heredity* 62:53, 1971.]

CHALLENGE QUESTIONS

Section 3.2

*40. A geneticist discovers an obese mouse in his laboratory colony. He breeds this obese mouse with a normal mouse. All the F_1 mice from this cross are normal in size. When he interbreeds two F_1 mice, eight of the F_2 mice are normal in size and two are obese. The geneticist then intercrosses two of his obese mice, and he finds that all the progeny from this cross are obese. These results lead the geneticist to conclude that obesity in mice results from a recessive allele.

A second geneticist at a different university also discovers an obese mouse in her laboratory colony. She carries out the same crosses as the first geneticist and obtains the same results. She also concludes that obesity in mice results from a recessive allele. One day the two geneticists meet at a genetics conference, learn of each other's experiments, and decide to exchange mice. They both find that when they cross two obese mice from the different laboratories, all the offspring are normal; however, when they cross two obese mice from the same laboratory, all the offspring are obese. Explain their results.

41. Albinism is a recessive trait in humans (see the introduction to Chapter 1). A geneticist studies a series of families in which both parents have pigmentation and at least one child has albinism. The geneticist reasons that both parents in these families must be heterozygotes and that albinism should appear in ¼ of their children. To his surprise, the geneticist finds that the frequency of albinism among the children of these families is significantly greater than ¼. Can you think of an explanation for the higher-than-expected frequency of albinism among these families?

ACTIVE LEARNING: THINK-PAIR-SHARE QUESTIONS

Introduction

1. About 40% of Solomon Islanders carry a gene for blond hair, and yet only 5%–10% of these people actually have blond hair. Why is the proportion of people with blond hair only 5%–10% when so many people carry the genes for blond hair?
2. Why is knowing the genetic basis of a trait such as blond hair important? Why would scientists go to the trouble of investigate the genetic basis of blond hair in Solomon Islanders?
3. If a blond-haired person from northern Europe mated with a blond Solomon Islander, what proportion of their offspring would be expected to have blond hair? Explain your reasoning.

Section 3.1

4. Why was Mendel's success dependent on his studying characteristics that exhibit only two easily distinguished phenotypes, such as white versus gray seed coats and round versus wrinkled seeds? Would he have been less successful if he had instead studied traits such as seed weight or leaf length, which vary much more in their phenotypes? Explain your answer.

Section 3.2

5. Geneticists often carry out reciprocal crosses when they are studying the inheritance of traits. Why do geneticists use reciprocal crosses?
6. Red hair in humans is inherited as a recessive trait. Bill and Sarah both have black hair. They marry and have four children, three of whom have red hair. Bill says it isn't genetically possible for two black-haired people to have $\frac{3}{4}$ red-haired children, and he accuses Sarah of infidelity. Sarah asserts that Bill doesn't understand genetics and probability, and that it is possible for two black-haired people to have $\frac{3}{4}$ red-haired children. Who is correct, and why?

Section 3.3

7. Are Mendel's principles of segregation and independent assortment even relevant today in the age of genomics, when it is possible to sequence an organism's entire genome and determine all of its genetic information? Why is it important to study these principles, and how can they be used?

Section 3.4

8. In corn, purple kernels (*P*) are dominant to yellow kernels (*p*), and starchy kernels (*Su*) are dominant to sugary kernels (*su*). A corn plant grown from a purple, starchy kernel is crossed with a plant grown from a yellow, sugary kernel, and the following progeny (kernels) are produced:

Phenotype	Number
purple, starchy	150
purple, sugary	142
yellow, starchy	161
yellow, sugary	115

Formulate a hypothesis about the genotypes of the parents and offspring in this cross. Perform a chi-square goodness-of-fit test comparing the observed numbers of progeny with the numbers expected based on your genetic hypothesis. What conclusion can you draw based on the results of your chi-square test? Can you suggest an explanation for the observed results?

ACTIVE LEARNING: CONCEPT MAPPING EXERCISES

Section 3.1

1. Outline the relationships among important genetic terminology by creating a concept map that relates the following terms: phenotype, gene, homozygous, heterozygous, locus, allele, genotype.

Section 3.2

2. Using a concept map, compare and contrast the principle of segregation (Mendel's first law) and the principle of independent assortment (Mendel's second law).

Section 3.5

3. Create a concept map that illustrates the different modes of inheritance and at least two characteristics of each.