# Basic Principles of Heredity



**Blond hair occurs in 5%–10% of dark-skinned Solomon Islanders.** Research demonstrates that 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 has 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 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 cytosine (C) in their *TYRP1* gene.

Further research showed that blond hair in 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

**3.1 Map of the Solomon Islands.**

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**
- Why is knowing the genetic basis of a trait such as blond hair important? Why would scientists go to the trouble to investigate the genetic basis of blond hair in Solomon Islanders?
- 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.

This chapter is about the principles of heredity: how genes—such as the one for blond hair in 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) (**Figure 3.2**).

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 two years of study 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.



**3.2  Gregor Johann Mendel discovered the principles of heredity by experimenting with peas.** [James King-Holmes/Science Source.]

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 so successfully to his genetic experiments.

## 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 3.3**), which offered clear advantages for genetic investigation. The plant is easy to cultivate, and Mendel had the monastery garden and greenhouse at his disposal. Compared with some other plants, peas grow relatively rapidly, completing an entire generation in a single growing season. By today's standards, one generation per year seems frightfully
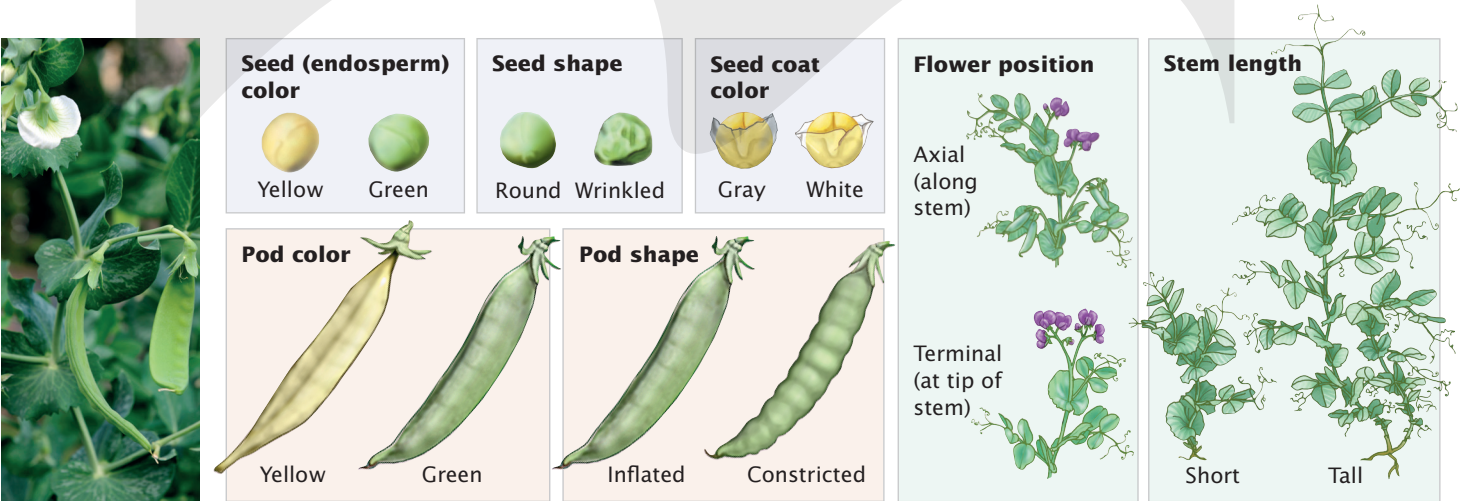


**3.3  Mendel used the pea plant *Pisum sativum* in his studies of heredity.** He examined seven characteristics that appeared in the seeds and in plants grown from the seeds. [Photograph by Charles Stirling/Alamy.]

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 that were available to Mendel were also crucial to his success because they differed in various traits and were genetically pure. Mendel was therefore able to begin with plants of variable and 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 3.3). He avoided characteristics that display a range of variation; instead, he focused his attention 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.

**THINK-PAIR-SHARE Question 2**

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 conducted additional crosses to test his hypotheses. He kept careful records of the numbers 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 13**

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**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 and his experimental approach.

**✔ 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

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## 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*

| TABLE 3.1 | Summary of important genetic terms |
|---|---|
| **Term** | **Definition** |
| Gene | An inherited factor (encoded in the 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 |
| Characteristic or character | An attribute or feature possessed by an organism |
| Phenotype or trait | The appearance or manifestation of a characteristic |

is a word that Mendel never knew. It was not coined until 1909, when Danish botanist Wilhelm Johannsen first used it. 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 3.4**). 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** for that gene. (The plural of locus is *loci*; it's bad form in genetics—and



Genes exist in different versions called alleles.

One allele encodes round seeds…

…and a different allele encodes wrinkled seeds.

Allele *R*          Allele *r*

Different alleles for a particular gene occupy the same locus on homologous chromosomes.
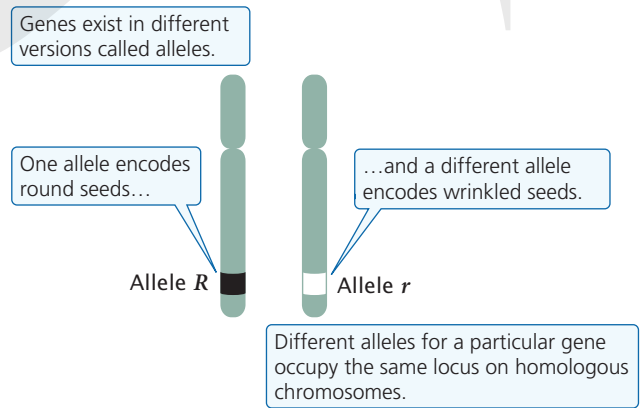
**3.4 At each locus, a diploid organism possesses two alleles located on different homologous chromosomes.** The alleles identified here refer to traits studied by Mendel.

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 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 genotype and phenotype?

## 3.2 Monohybrid Crosses Reveal the Principle of Segregation and the Concept of Dominance
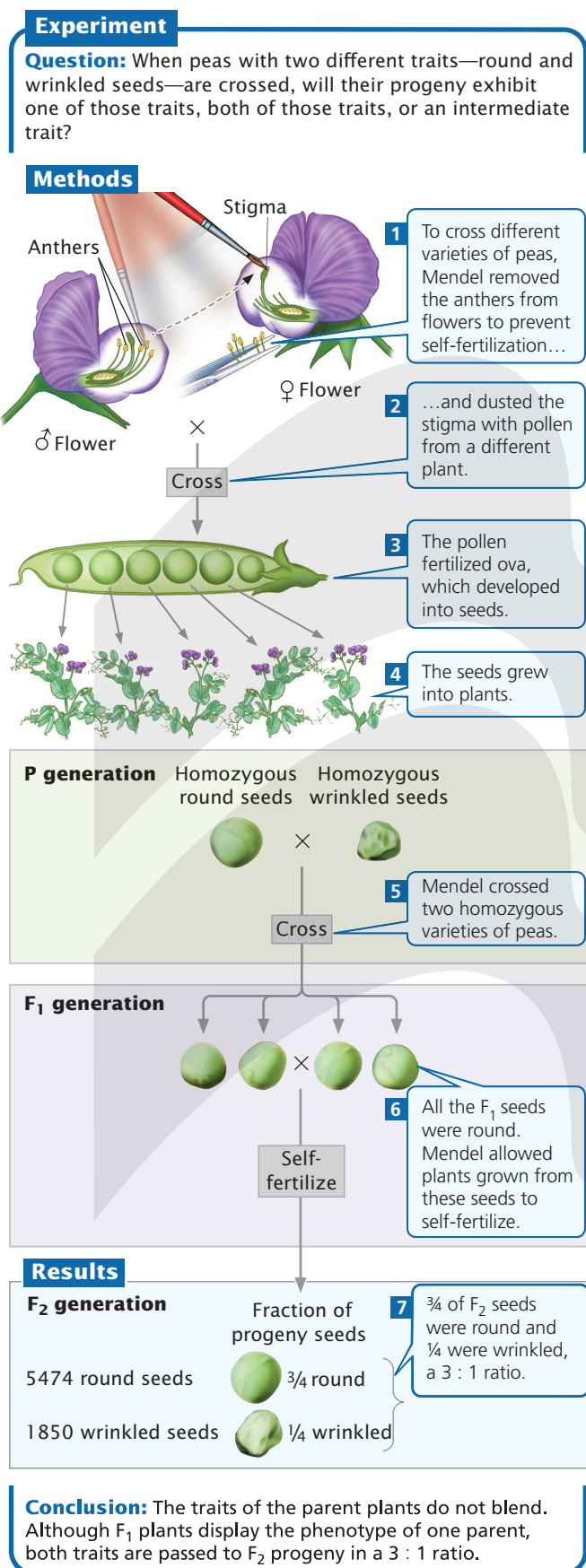
Mendel started with 34 varieties of peas and spent two years selecting those varieties that he would use in his experiments. He verified that each variety was pure-breeding (homozygous for each of the traits that he chose to study) by growing the plants for two generations and confirming that all offspring were the same as their parents. He then carried out a number of crosses between the different varieties. Although peas are normally self-fertilizing (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 3.5**).

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 3.5). 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 3**

## Experiment

**Question:** When peas with two different traits—round and wrinkled seeds—are crossed, will their progeny exhibit one of those traits, both of those traits, or an intermediate trait?

### Methods

Stigma

Anthers

♂ Flower   ♀ Flower

Cross

**1** To cross different varieties of peas, Mendel removed the anthers from flowers to prevent self-fertilization…

**2** …and dusted the stigma with pollen from a different plant.

**3** The pollen fertilized ova, which developed into seeds.

**4** The seeds grew into plants.

**P generation**   Homozygous round seeds   Homozygous wrinkled seeds

Cross

**5** Mendel crossed two homozygous varieties of peas.

**F₁ generation**

Self-fertilize

**6** All the $F_1$ seeds were round. Mendel allowed plants grown from these seeds to self-fertilize.

### Results

**F₂ generation**   Fraction of progeny seeds

**7** ¾ of $F_2$ seeds were round and ¼ were wrinkled, a 3 : 1 ratio.

5474 round seeds   ¾ round

1850 wrinkled seeds   ¼ wrinkled

**Conclusion:** The traits of the parent plants do not blend. Although $F_1$ plants display the phenotype of one parent, both traits are passed to $F_2$ progeny in a 3 : 1 ratio.

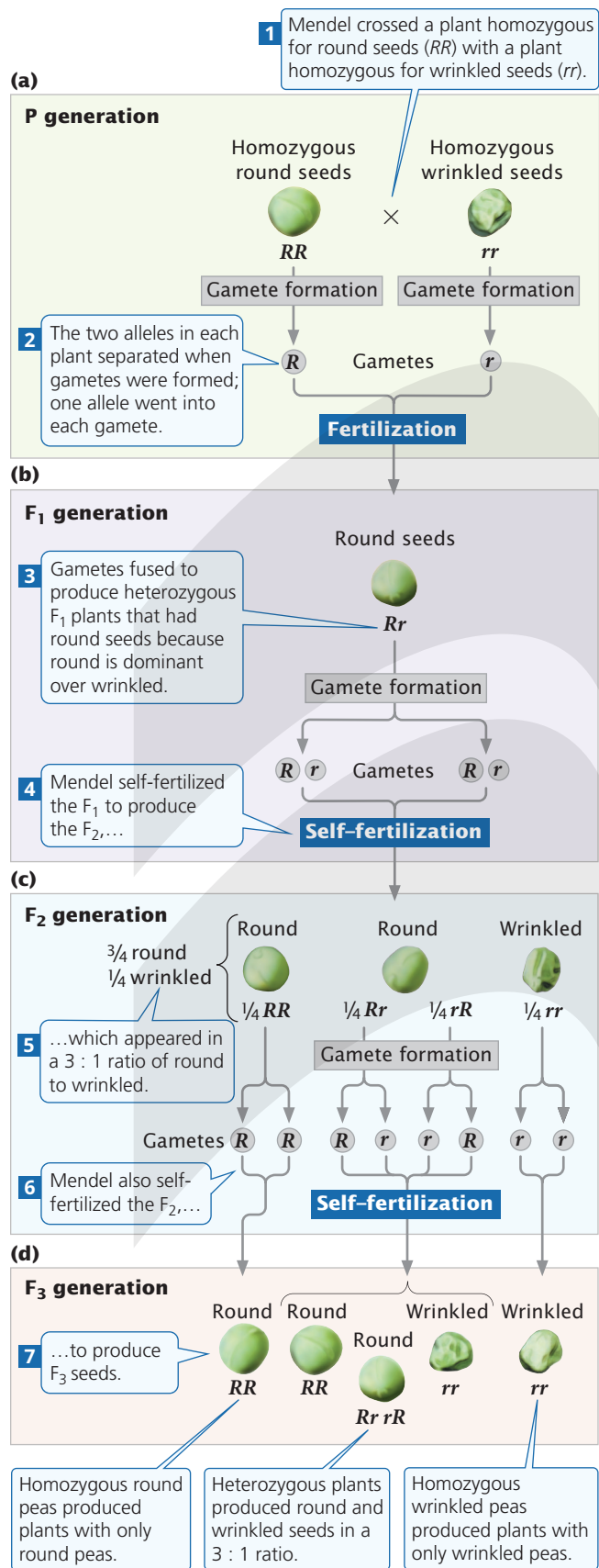**3.5 Mendel conducted monohybrid crosses.**

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 3.5). He noticed that the numbers of the round and wrinkled seeds constituted approximately a 3 to 1 ratio; that is, about ¾ of the $F_2$ seeds were round and ¼ 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 of 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.

## 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 3.6a**).

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 3.6b**). 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,

**(a)**

**P generation**

1 Mendel crossed a plant homozygous for round seeds (*RR*) with a plant homozygous for wrinkled seeds (*rr*).

Homozygous round seeds

Homozygous wrinkled seeds

×

*RR*

*rr*

Gamete formation | Gamete formation

2 The two alleles in each plant separated when gametes were formed; one allele went into each gamete.

*R*    Gametes    *r*

**Fertilization**

**(b)**

**F₁ generation**

Round seeds

*Rr*

3 Gametes fused to produce heterozygous F₁ plants that had round seeds because round is dominant over wrinkled.

Gamete formation

*R* *r*    Gametes    *R* *r*

4 Mendel self-fertilized the F₁ to produce the F₂,…

**Self–fertilization**

**(c)**

**F₂ generation**

¾ round
¼ wrinkled

Round    Round    Wrinkled

¼ *RR*    ¼ *Rr*    ¼ *rR*    ¼ *rr*

5 …which appeared in a 3 : 1 ratio of round to wrinkled.

Gamete formation

Gametes *R* *R*    *R* *r*    *r* *R*    *r* *r*

6 Mendel also self-fertilized the F₂,…

**Self–fertilization**

**(d)**

**F₃ generation**

Round  Round        Wrinkled  Wrinkled
        Round

7 …to produce F₃ seeds.

*RR*    *RR*    *rr*    *rr*
        *Rr rR*

Homozygous round peas produced plants with only round peas.

Heterozygous plants produced round and wrinkled seeds in a 3 : 1 ratio.

Homozygous wrinkled peas produced plants with only wrinkled peas.

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

the effect of the recessive allele is not observed in the phenotype. The 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 F₁ (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 F₂: *RR*, *Rr*, *rR*, *rr* (**Figure 3.6c**). Because round (*R*) is dominant to wrinkled (*r*), there were three round-seeded progeny (*RR*, *Rr*, *rR*) for every wrinkled-seeded progeny (*rr*) in the F₂. This 3 : 1 ratio of round-seeded to wrinkled-seeded progeny that Mendel observed in the F₂ could be obtained only if the two alleles of a genotype separated into the gametes with equal probability.

The conclusions that Mendel drew about inheritance from his monohybrid crosses have been further developed and formalized into the principle of segregation and the concept of dominance. The **principle of segregation** (**Mendel's first law**; see **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.

| 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.

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 3.6d**).

The plants grown from round seeds—the dominant trait—fell into two types (see Figure 3.6c). Following 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 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 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 water. Because *rr* seeds absorb more water and expand more during development, they lose more water during maturation and afterward appear 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 18.
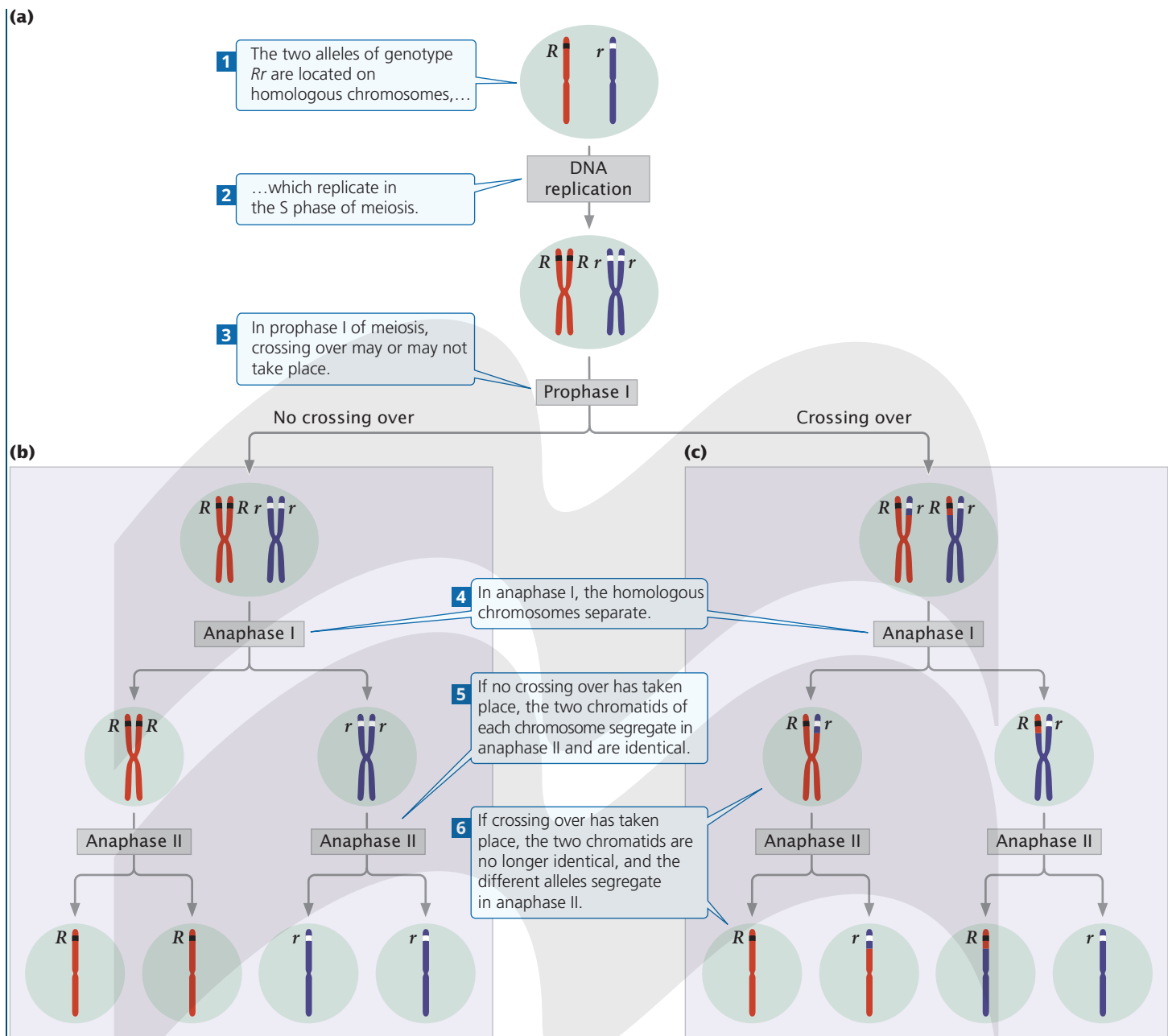
> ## CONNECTING CONCEPTS  CONNECT
>
> ### Relating Genetic Crosses to Meiosis
>
> We have now seen how the results of monohybrid crosses are explained by Mendel's principle of segregation. Many students find that they enjoy working genetic crosses but are frustrated by the abstract nature of the symbols. 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.

**(a)**

1  The two alleles of genotype *Rr* are located on homologous chromosomes,…

*R    r*

DNA replication

2  …which replicate in the S phase of meiosis.

*R   R r    r*

3  In prophase I of meiosis, crossing over may or may not take place.

Prophase I

No crossing over                                    Crossing over

**(b)**                                             **(c)**

*R   R r    r*                                      *R   r R    r*

4  In anaphase I, the homologous chromosomes separate.

Anaphase I                                          Anaphase I

*R    R*          *r    r*                           *R    r*          *R    r*

5  If no crossing over has taken place, the two chromatids of each chromosome segregate in anaphase II and are identical.

Anaphase II       Anaphase II                        Anaphase II       Anaphase II

6  If crossing over has taken place, the two chromatids are no longer identical, and the different alleles segregate in anaphase II.

*R*      *R*      *r*      *r*                        *R*      *r*      *R*      *r*

**3.7  Segregation results from the separation of homologous chromosomes in meiosis.**

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 3.7a**). The homologous chromosomes segregate in anaphase I, thereby separating the two different alleles (**Figure 3.7b and c**). 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 (see Figure 3.7c).

Mendel knew nothing 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 30**

## Predicting the Outcomes of Genetic Crosses

One of Mendel's goals in conducting his experiments in 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.
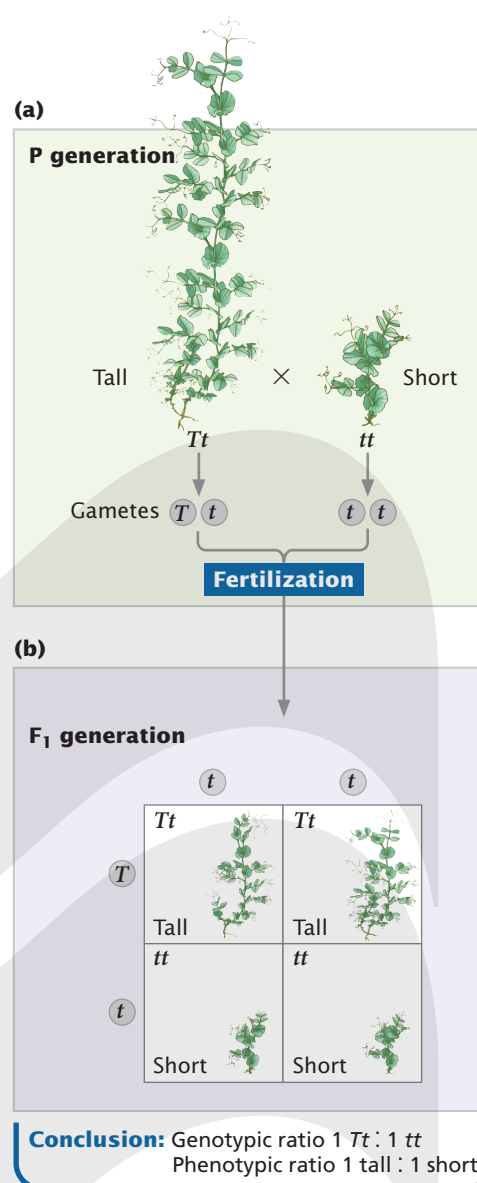
**THE PUNNETT SQUARE** The Punnett square was developed by the English geneticist Reginald C. Punnett in 1917. To illustrate the Punnett square, let's examine another cross carried out by Mendel. By crossing two varieties of pea plants that differed in height, Mendel established that tall ($T$) was dominant 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 3.8a**). 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 ($Tt$), so 50% of its gametes will receive a tall allele ($T$) and the other 50% will receive a short allele ($t$).

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 3.8b**). 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 3.8b, 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 3.8b, 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 $Tt$ (and the tall phenotype) and ½ of the progeny to have genotype $tt$ (and 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 3.6b 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 $rr$).



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

### 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 3.6b 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 $\frac{1}{6}$ 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.
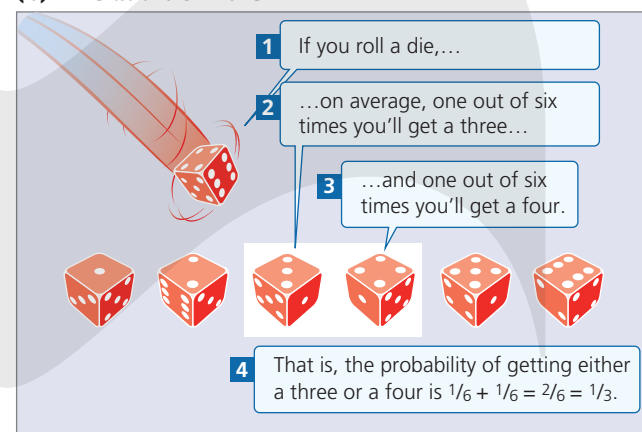
To illustrate the use of the multiplication rule, let's again consider the roll of a die. The probability of rolling one die and obtaining a four is $\frac{1}{6}$. To calculate the probability of rolling a die twice and obtaining two fours, we can apply the multiplication rule. The probability of obtaining 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 3.9a**). 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 are rusty and need a review, turn to the appendix at the end of the book entitled Working with Fractions [pp. B1–B3].)

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

**(a) The multiplication rule**



1  If you roll a die,…

2  …in a large number of sample rolls, on average, one out of six times you will obtain a four;…

**Roll 1**

3  …so the probability of obtaining a four in any roll is $1/6$.

4  If you roll the die again,…

5  …your probability of getting four is again $1/6$;…

**Roll 2**

6  …so the probability of getting a four on the first roll and the second roll is $1/6 \times 1/6 = 1/36$.

**(b) The addition rule**



1  If you roll a die,…

2  …on average, one out of six times you'll get a three…

3  …and one out of six times you'll get a four.

4  That is, the probability of getting either a three or a four is $1/6 + 1/6 = 2/6 = 1/3$.

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

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 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 $\frac{1}{6} + \frac{1}{6} = \frac{2}{6} = \frac{1}{3}$ (**Figure 3.9b**). 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 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 receive a $t$ allele from the first parent and a $T$ allele from the second.

After determining the probabilities 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 $tT$, so, 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 $\frac{2}{3}$.

**THE BINOMIAL EXPANSION AND PROBABILITY** When probability is used, it is important to recognize that there may be several different ways in which a set of events can occur. Consider two parents who are both heterozygous for albinism, a recessive condition in humans that causes reduced pigmentation in the skin, hair, and eyes (**Figure 3.10**; see also the introduction to Chapter 1). When two parents heterozygous for albinism mate ($Aa \times Aa$), the probability of their having a child with albinism ($aa$) is $\frac{1}{4}$, and the probability of their having a child with full pigmentation ($AA$ or $Aa$) is $\frac{3}{4}$. Suppose we want to know the probability of this couple having three children with albinism.
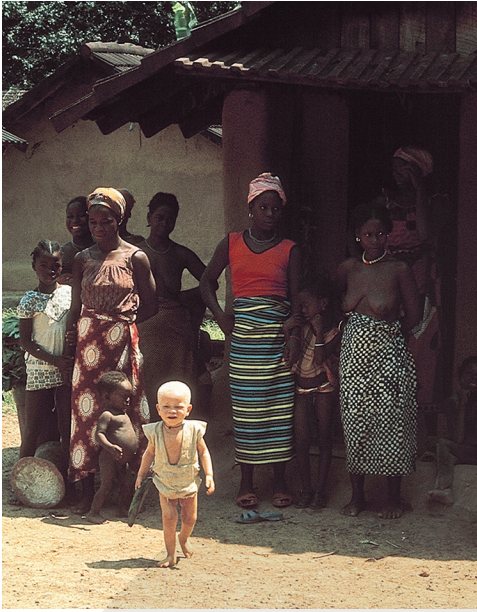
**3.10 Albinism in humans is usually inherited as a recessive trait.**
[Richard Hutchings/Science Source.]

In this case, there is only one way in which this can happen: their first child has albinism *and* their second child has albinism *and* their third child has albinism. Here, we simply apply the multiplication rule: $\frac{1}{4} \times \frac{1}{4} \times \frac{1}{4} = \frac{1}{64}$.

Suppose we now ask what the probability is of this couple having three children, one with albinism and two with pigmentation. This situation is more complicated. The first child might have albinism, whereas the second and third might be unaffected; the probability of this sequence of events is $\frac{1}{4} \times \frac{3}{4} \times \frac{3}{4} = \frac{9}{64}$. Alternatively, the first and third children might have pigmentation, whereas the second might have albinism; the probability of this sequence is $\frac{3}{4} \times \frac{1}{4} \times \frac{3}{4} = \frac{9}{64}$. Finally, the first two children might have pigmentation and the third albinism; the probability of this sequence is $\frac{3}{4} \times \frac{3}{4} \times \frac{1}{4} = \frac{9}{64}$. Because *either* the first sequence *or* the second sequence *or* the third sequence produces one child with albinism and two with pigmentation, we apply the addition rule and add the probabilities: $\frac{9}{64} + \frac{9}{64} + \frac{9}{64} = \frac{27}{64}$.

If we want to know the probability of this couple having five children, two with albinism and three with pigmentation, figuring out *all* the different combinations of children and their probabilities becomes more difficult. This task is made easier if we apply the binomial expansion.

The binomial takes the form $(p + q)^n$, where $p$ equals the probability of one event, $q$ equals the probability of the alternative event, and $n$ equals the number of times the event occurs. For figuring the probability of two out of five children with albinism,

$p =$ the probability of a child having albinism ($\frac{1}{4}$)

$q =$ the probability of a child having pigmentation ($\frac{3}{4}$)

The binomial for this situation is $(p + q)^5$ because there are five children in the family ($n = 5$). The expansion of this binomial is

$$(p + q)^5 = p^5 + 5p^4q + 10p^3q^2 + 10p^2q^3 + 5pq^4 + q^5$$

Each of the terms in the expansion provides the probability of one particular combination of traits in the children. The first term in the expansion ($p^5$) equals the probability of having five children with albinism because $p$ is the probability of albinism. The second term ($5p^4q$) equals the probability of having four children with albinism and one with pigmentation, the third term ($10p^3q^2$) equals the probability of having three children with albinism and two with pigmentation, and so forth.

To obtain the probability of any combination of events, we insert the values of $p$ and $q$. Thus, the probability of having two out of five children with albinism is

$$10p^2q^3 = 10\left(\tfrac{1}{4}\right)^2 \left(\tfrac{3}{4}\right)^3 = \tfrac{270}{1024} = 0.26$$

We can easily figure out the probability of any desired combination of albinism and pigmentation among five children by using the other terms in the expansion.

How did we expand the binomial in this example? In general, the expansion of any binomial $(p + q)^n$ consists of a series of $n + 1$ terms. In the preceding example, $n = 5$, so there are $5 + 1 = 6$ terms: $p^5$, $5p^4q$, $10p^3q^2$, $10p^2q^3$, $5pq^4$, and $q^5$. To write out the terms, first figure out their exponents. The exponent of $p$ in the first term is always the power to which the binomial is raised, or $n$. In our example, $n = 5$, so our first term is $p^5$. The exponent of $p$ decreases by one in each successive term, so the exponent of $p$ is 4 in the second term ($p^4$), 3 in the third term ($p^3$), and so forth. The exponent of $q$ is 0 (no $q$) in the first term and increases by one in each successive term, increasing from 0 to 5 in our example.

Next, we determine the coefficient of each term. The coefficient of the first term is always 1, so in our example, the first term is $1p^5$, or just $p^5$. The coefficient of the second term is always the same as the power to which the binomial is raised; in our example, this coefficient is 5, and the term is $5p^4q$. For the coefficient of the third term, we look back at the preceding term, multiply the coefficient of the preceding term (5 in our example) by the exponent of $p$ in that term (4), and then divide by the number of that term (second term, or 2). So the coefficient of the third term in our example is $(5 \times 4) / 2 = \frac{20}{2} = 10$, and the term is $10p^3q^2$. We follow this procedure for each successive term. The coefficients for the terms in the binomial expansion can also be determined from Pascal's triangle (**Table 3.3**). The exponents and coefficients for each term in the first five binomial expansions are given in **Table 3.4**.

Another way to determine the probability of any particular combination of events is to use the following formula:

$$P = \frac{n!}{s!t!} p^s q^t$$

where $P$ equals the overall probability of event X with probability $p$ occurring $s$ times and alternative event Y with

**TABLE 3.3 Pascal's triangle**

The numbers in each row represent the coefficients of each term in the binomial expansion $(p + q)^n$.

| $n$ | Coefficients | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | | | | | | 1 | | | | | | | |
| 1 | | | | | 1 | | 1 | | | | | |
| 2 | | | | 1 | | 2 | | 1 | | | | |
| 3 | | | 1 | | 3 | | 3 | | 1 | | | |
| 4 | | 1 | | 4 | | 6 | | 4 | | 1 | | |
| 5 | 1 | | 5 | | 10 | | 10 | | 5 | | 1 | |
| 6 | 1 | | 6 | 15 | | 20 | | 15 | | 6 | | 1 |

*Note:* Each number in the triangle, except for the first, is equal to the sum of the two numbers directly above it.

**TABLE 3.4 Coefficients and terms for the binomial expansion $(p + q)^n$ for $n = 1$ through 5**

| $n$ | Binomial Expansion |
|---|---|
| 1 | $a + b$ |
| 2 | $a^2 + 2ab + b^2$ |
| 3 | $a^3 + 3a^2b + 3ab^2 + b^3$ |
| 4 | $a^4 + 4a^3b + 6a^2b^2 + 4ab^3 + b^4$ |
| 5 | $a^5 + 5a^4b + 10a^3b^2 + 10a^2b^3 + 5ab^4 + b^5$ |

probability $q$ occurring $t$ times. For our albinism example, event X would be the occurrence of a child with albinism ($\frac{1}{4}$) and event Y would be the occurrence of a child with pigmentation ($\frac{3}{4}$); $s$ would equal the number of children with albinism (2) and $t$ would equal the number of children with pigmentation (3). The ! symbol stands for factorial, and it means the product of all the integers from $n$ to 1. In this example, $n = 5$; so $n! = 5 \times 4 \times 3 \times 2 \times 1$. Applying this formula to obtain the probability of two out of five children having albinism, we obtain

$$P = \frac{5!}{2!3!}(\tfrac{1}{4})^2(\tfrac{3}{4})^3 = \frac{5 \times 4 \times 3 \times 2 \times 1}{2 \times 1 \times 3 \times 2 \times 1}(\tfrac{1}{4})^2(\tfrac{3}{4})^3 = 0.26$$

This value is the same as that obtained with the binomial expansion. **> TRY PROBLEMS 25, 26, AND 27**

**THINK-PAIR-SHARE Question 4**

## 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 3.8 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 PROBLEMS 18 AND 21**

**THINK-PAIR-SHARE Question 5**

**CONCEPTS**

The binomial expansion can be used to determine the probability of a particular set of events. 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 the 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 written $El^+/El^R$, 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   CONNECT

#### 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 alleles is dominant to the other. 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 use these ratios to work more complicated crosses that include several loci.

There are only three phenotypic ratios to understand (**Table 3.5**). The first is the 3 : 1 ratio, which arises in a simple genetic cross when both of the parents are heterozygous for a dominant trait ($Aa \times Aa$). The second phenotypic ratio is the 1 : 1 ratio, which results from the mating of a heterozygous parent and a homozygous recessive parent. The homozygous parent in this cross must carry two recessive alleles ($Aa \times aa$) because a cross between a homozygous dominant parent and a heterozygous parent ($AA \times Aa$) produces offspring displaying only the dominant trait.

| TABLE 3.5 | Phenotypic ratios for simple genetic crosses (crosses for a single locus) with dominance |
|---|---|

An underline in a genotype, such as $A\_$, indicates that any allele is possible.

| Phenotypic Ratio | Genotypes of Parents | Genotypes of Progeny |
|---|---|---|
| 3 : 1 | $Aa \times Aa$ | $\frac{3}{4}\ A\_ : \frac{1}{4}\ aa$ |
| 1 : 1 | $Aa \times aa$ | $\frac{1}{2}\ Aa : \frac{1}{2}\ aa$ |
| Uniform progeny | $AA \times AA$ | All $AA$ |
| | $aa \times aa$ | All $aa$ |
| | $AA \times aa$ | All $Aa$ |
| | $AA \times Aa$ | All $A\_$ |

The third phenotypic ratio is not really a ratio: all the offspring have the same phenotype (uniform progeny). Several combinations of parental genotypes can produce this outcome (see Table 3.5). A cross between any two homozygous parents—either between two parents of the same homozygous genotype ($AA \times AA$ or $aa \times aa$) or between two parents with different homozygous genotypes ($AA \times aa$)—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 ($AA \times Aa$).

If we are interested in the ratios of genotypes instead of phenotypes, there are still only three outcomes to remember (**Table 3.6**): 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 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 not only for a single locus but also, as you will see in the next section, for multiple loci.

| TABLE 3.6 | Genotypic ratios for simple genetic crosses (crosses for a single locus) |
|---|---|

| Genotypic Ratio | Genotypes of Parents | Genotypes of Progeny |
|---|---|---|
| 1 : 2 : 1 | $Aa \times Aa$ | $\frac{1}{4}\ AA : \frac{1}{2}\ Aa : \frac{1}{4}\ aa$ |
| 1 : 1 | $Aa \times aa$ | $\frac{1}{2}\ Aa : \frac{1}{2}\ aa$ |
| | $Aa \times AA$ | $\frac{1}{2}\ Aa : \frac{1}{2}\ AA$ |
| Uniform progeny | $AA \times AA$ | All $AA$ |
| | $aa \times aa$ | All $aa$ |
| | $AA \times aa$ | 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 3.11a**). The principle of segregation tells us that the alleles at each locus separate, and that one allele for each locus passes into 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.
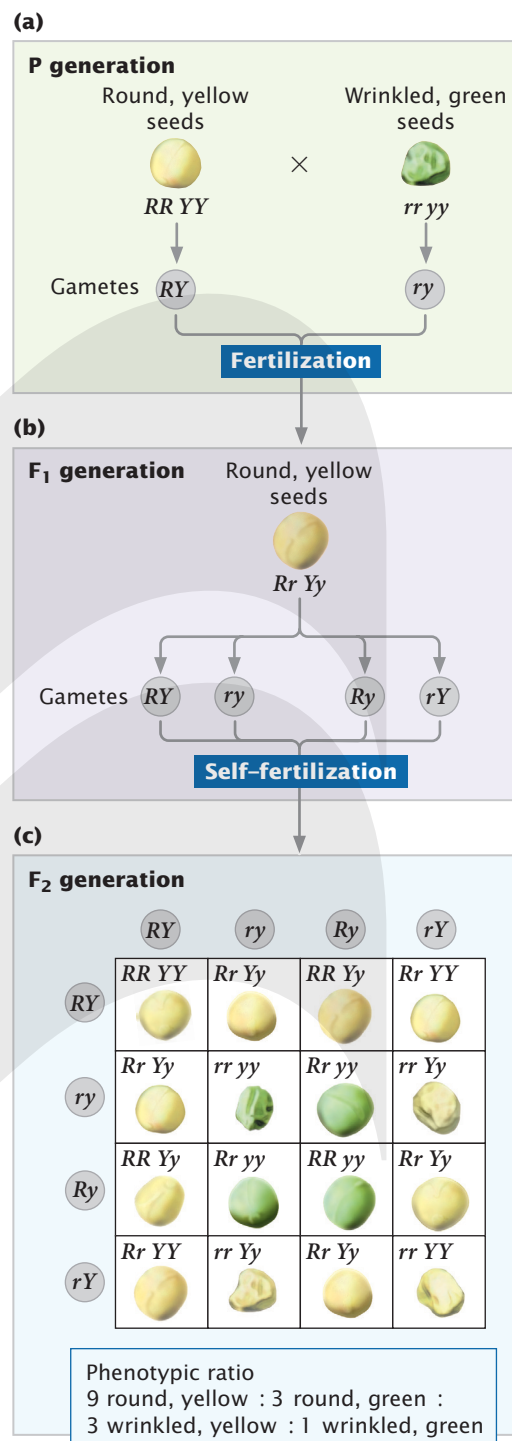
When Mendel allowed the $F_1$ plants to self-fertilize to produce the $F_2$, the alleles at each locus separated, and one of those alleles passed into each gamete. This event is where the principle of independent assortment becomes important. The two pairs of alleles can separate in two ways: (1) *R* 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 3.11b**). When these four types of gametes are combined to produce the $F_2$ generation, the progeny consist of $\frac{9}{16}$ round and yellow, $\frac{3}{16}$ round and green, $\frac{3}{16}$ wrinkled and yellow, and $\frac{1}{16}$ wrinkled and green, resulting in a 9 : 3 : 3 : 1 phenotypic ratio (**Figure 3.11c**).

**THINK-PAIR-SHARE Question 6**



**Experiment**

**Question:** Do alleles encoding different traits separate independently?

**(a)**

**Methods**

**P generation**

Round, yellow seeds    Wrinkled, green seeds

*RR YY*    ×    *rr yy*

Gametes   *RY*         *ry*

**Fertilization**

**(b)**

**F₁ generation**    Round, yellow seeds

*Rr Yy*

Gametes   *RY*   *ry*   *Ry*   *rY*

**Self–fertilization**

**(c)**

**Results**

**F₂ generation**

|  | *RY* | *ry* | *Ry* | *rY* |
|---|---|---|---|---|
| *RY* | *RR YY* | *Rr Yy* | *RR Yy* | *Rr YY* |
| *ry* | *Rr Yy* | *rr yy* | *Rr yy* | *rr Yy* |
| *Ry* | *RR Yy* | *Rr yy* | *RR yy* | *Rr Yy* |
| *rY* | *Rr YY* | *rr Yy* | *Rr Yy* | *rr YY* |

Phenotypic ratio
9 round, yellow : 3 round, green : 3 wrinkled, yellow : 1 wrinkled, green

**Conclusion:** The allele encoding color separated independently of the allele encoding seed shape, producing a 9 : 3 : 3 : 1 ratio in the $F_2$ progeny.
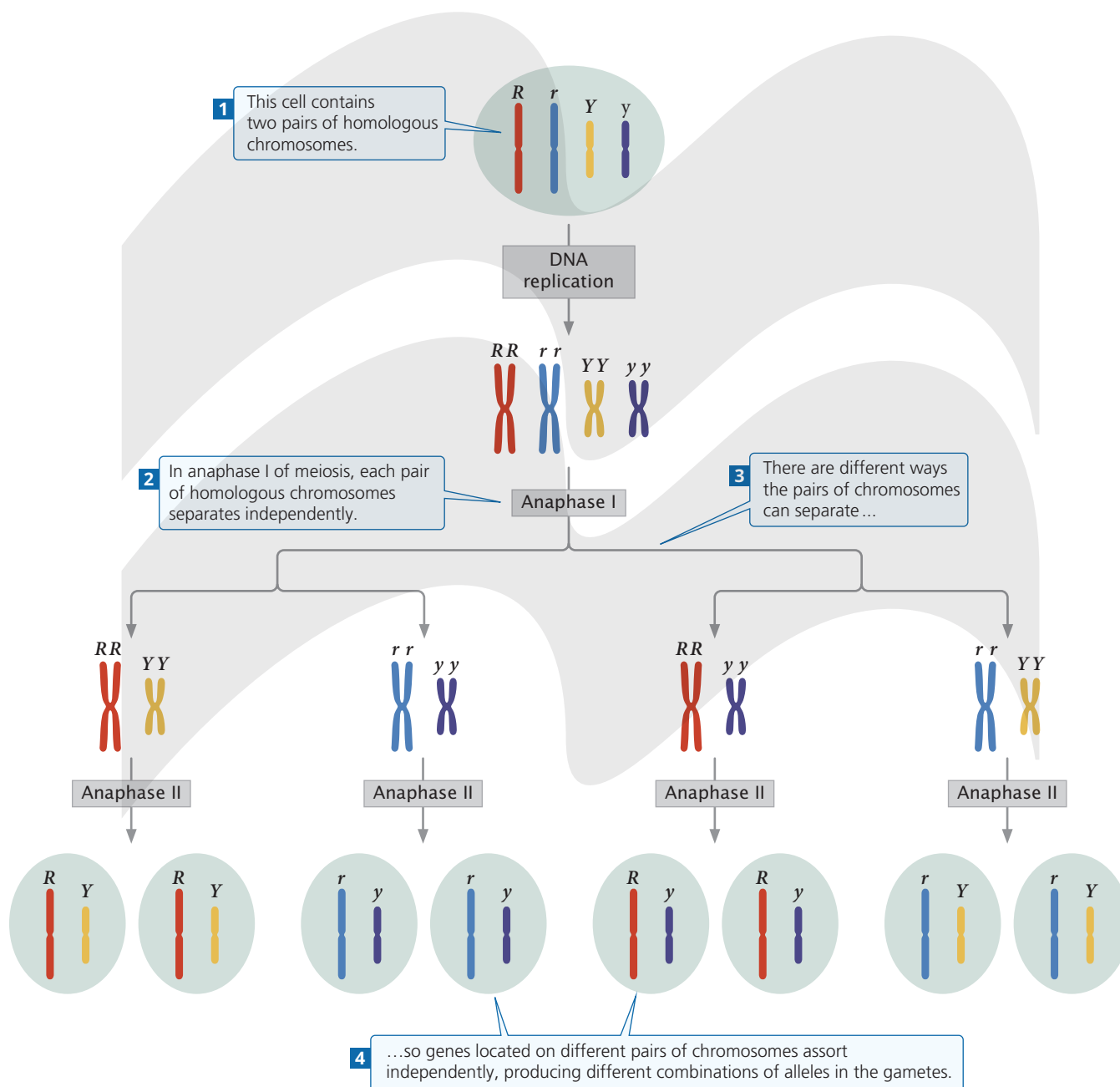
**3.11  Mendel's dihybrid crosses revealed the principle of independent assortment.**

## 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 3.12**), so genes located on different pairs

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 Chapter 7).



**3.12  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 do not, however, assort independently.

**✔ CONCEPT CHECK 6**

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

## 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 ($Rr\ Yy \times Rr\ Yy$) by considering each characteristic separately (**Figure 3.13a**). If we consider only the shape of the seeds, the cross was $Rr \times Rr$, which yields a 3 : 1 phenotypic ratio ($\frac{3}{4}$ round and $\frac{1}{4}$ wrinkled progeny; see Table 3.5). Next, we consider the other characteristic, the color of the seeds. The cross was $Yy \times Yy$, which produces a 3 : 1 phenotypic ratio ($\frac{3}{4}$ yellow and $\frac{1}{4}$ 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 $\frac{3}{4}$ (the probability of round) $\times \frac{3}{4}$ (the probability of yellow) $= \frac{9}{16}$. The proportion of progeny with round and green seeds is $\frac{3}{4} \times \frac{1}{4} = \frac{3}{16}$; the proportion of progeny with wrinkled and yellow seeds is $\frac{1}{4} \times \frac{3}{4} = \frac{3}{16}$; and the proportion of progeny with wrinkled and green seeds is $\frac{1}{4} \times \frac{1}{4} = \frac{1}{16}$.

Branch diagrams are a convenient way of organizing all the combinations of characteristics in the progeny of a cross (**Figure 3.13b**). In the first column, list the proportions of the phenotypes for one characteristic (here, seed shape: $\frac{3}{4}$ round and $\frac{1}{4}$ wrinkled). In the second column, list the proportions of the phenotypes for the next characteristic (seed color: $\frac{3}{4}$ yellow and $\frac{1}{4}$ green) twice, next to each of the phenotypes in the first column: write "$\frac{3}{4}$ yellow" and "$\frac{1}{4}$ 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., $\frac{3}{4}$ yellow and $\frac{1}{4}$ 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 ($\frac{3}{4}$) and yellow ($\frac{3}{4}$) seeds is $\frac{3}{4} \times \frac{3}{4} = \frac{9}{16}$. The advantage of the branch diagram is that it helps us keep track of all the potential combinations of traits that may
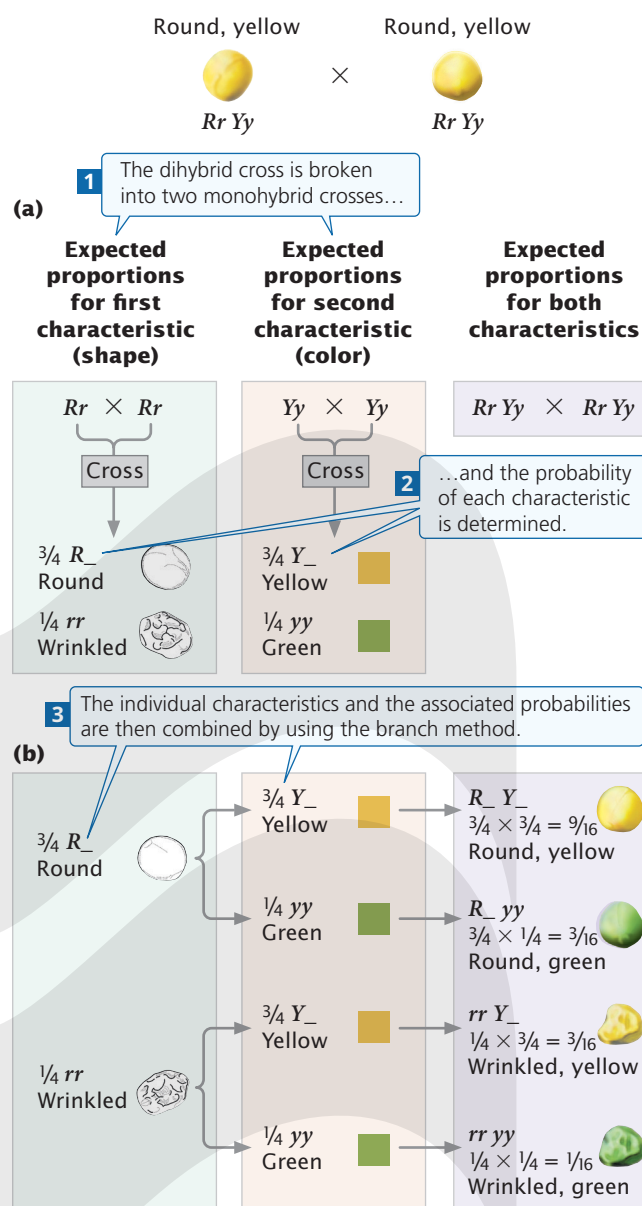


**3.13 A branch diagram can be used to determine the phenotypes and expected proportions of offspring from a dihybrid cross ($Rr\ Yy \times Rr\ Yy$).**

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 3.5 and 3.6. 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 3.11. The probability of obtaining the $Rr$ genotype in a cross of $Rr \times Rr$ is $\frac{1}{2}$, and that of obtaining the $yy$ genotype in a cross of $Yy \times Yy$ is $\frac{1}{4}$ (see Table 3.6). Using the multiplication rule, we find the probability of $Rr\ yy$ to be $\frac{1}{2} \times \frac{1}{4} = \frac{1}{8}$.

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* | $\frac{1}{4}$ |
| *Bb × Bb* | *bb* | $\frac{1}{4}$ |
| *cc × Cc* | *cc* | $\frac{1}{2}$ |
| *Dd × dd* | *dd* | $\frac{1}{2}$ |
| *Ee × Ee* | *ee* | $\frac{1}{4}$ |

The probability of an offspring from this cross having genotype *aa bb cc dd ee* is now easily obtained by using the multiplication rule: $\frac{1}{4} \times \frac{1}{4} \times \frac{1}{2} \times \frac{1}{2} \times \frac{1}{4} = \frac{1}{256}$. This calculation assumes that the genes at these five loci all 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**.

**Ⓐ**

> **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).

## 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 3.14**. First, break the cross down into a series of single-locus crosses. The cross *Rr × rr* yields $\frac{1}{2}$ round (*Rr*) progeny and $\frac{1}{2}$ wrinkled (*rr*) progeny. The cross *Yy × yy* yields $\frac{1}{2}$ yellow (*Yy*) progeny and $\frac{1}{2}$ green (*yy*) progeny. Using the multiplication rule, we find the proportion of round and yellow progeny to be $\frac{1}{2}$ (the probability of round) $\times \frac{1}{2}$ (the probability of yellow) $= \frac{1}{4}$. Four combinations of traits appear in the offspring in the following proportions: $\frac{1}{4}$ *Rr Yy*, round and yellow; $\frac{1}{4}$ *Rr yy*, round and green; $\frac{1}{4}$ *rr Yy*, wrinkled and yellow; and $\frac{1}{4}$ *rr yy*, wrinkled and green.

**THINK-PAIR-SHARE Question 7**
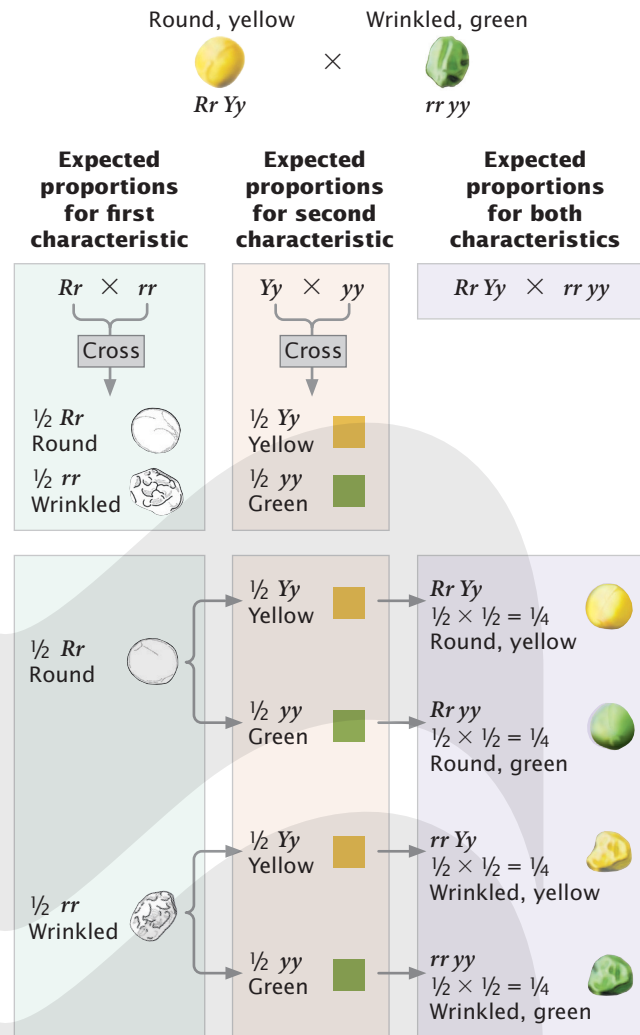


**3.14 A branch diagram can be used to determine the phenotypes and expected proportions of offspring from a dihybrid testcross (*Rr Yy × rr yy*).**

> **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, for 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:

$$B—\text{black} \qquad S—\text{solid}$$
$$b—\text{brown} \qquad s—\text{spotted}$$

Next, write out the crosses given in the problem:

| P | Homozygous black, spotted | $\times$ | Homozygous brown, solid |
|---|---|---|---|

$F_1$                Black, solid

Testcross    Black, solid    $\times$    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* | $\times$ | Homozygous brown, solid *bb SS* |
|---|---|---|---|

$F_1$              Black, solid *B_ S_*

Testcross    Black, solid *B_ S_*    $\times$    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*:

| P | Homozygous black, spotted *BB ss* | $\times$ | Homozygous brown, solid *bb SS* |
|---|---|---|---|

Gametes      (*Bs*)          (*bS*)

$F_1$               *Bb Ss*

Use the $F_1$ genotype to work the testcross ($Bb\,Ss \times bb\,ss$), breaking it into two single-locus crosses. First, consider the cross for coat color: $Bb \times bb$. Any cross between a heterozygote and a homozygous recessive genotype produces a 1 : 1 phenotypic ratio of progeny (see Table 3.5):

$$Bb \times bb$$
$$\downarrow$$
$\frac{1}{2}$ $Bb$ black
$\frac{1}{2}$ $bb$ brown

Next, consider the cross for spotting: $Ss \times ss$. This cross is also between a heterozygote and a homozygous recessive genotype and produces $\frac{1}{2}$ solid ($Ss$) and $\frac{1}{2}$ spotted ($ss$) progeny (see Table 3.5).

$$Ss \times ss$$
$$\downarrow$$
$\frac{1}{2}$ $Ss$ solid
$\frac{1}{2}$ $ss$ spotted

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

$\frac{1}{2}$ $Bb$ black
— $\frac{1}{2}$ $Ss$ solid $\longrightarrow$ $Bb\,Ss$ black, solid $\frac{1}{2} \times \frac{1}{2} = \frac{1}{4}$
— $\frac{1}{2}$ $ss$ spotted $\longrightarrow$ $Bb\,ss$ black, spotted $\frac{1}{2} \times \frac{1}{2} = \frac{1}{4}$

$\frac{1}{2}$ $bb$ brown
— $\frac{1}{2}$ $Ss$ solid $\longrightarrow$ $bb\,Ss$ brown, solid $\frac{1}{2} \times \frac{1}{2} = \frac{1}{4}$
— $\frac{1}{2}$ $ss$ spotted $\longrightarrow$ $bb\,ss$ brown, spotted $\frac{1}{2} \times \frac{1}{2} = \frac{1}{4}$

**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 33** at the end of this 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—$\frac{1}{2}$ heads and $\frac{1}{2}$ 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 ($\chi^2$) 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 ($\chi^2$) value is calculated by using the following formula:

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

where $\Sigma$ 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 (observed − expected)$^2$/ 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 degrees of freedom are equal to $n - 1$, in which

**TABLE 3.7**    **Critical values of the $\chi^2$ distribution**

| df | | | | | | *P* | | | |
|---|---|---|---|---|---|---|---|---|---|
| | **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 the observed and the expected values in a chi-square test.

$n$ 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 3.7**). 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 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 3.15.** > TRY PROBLEM 38
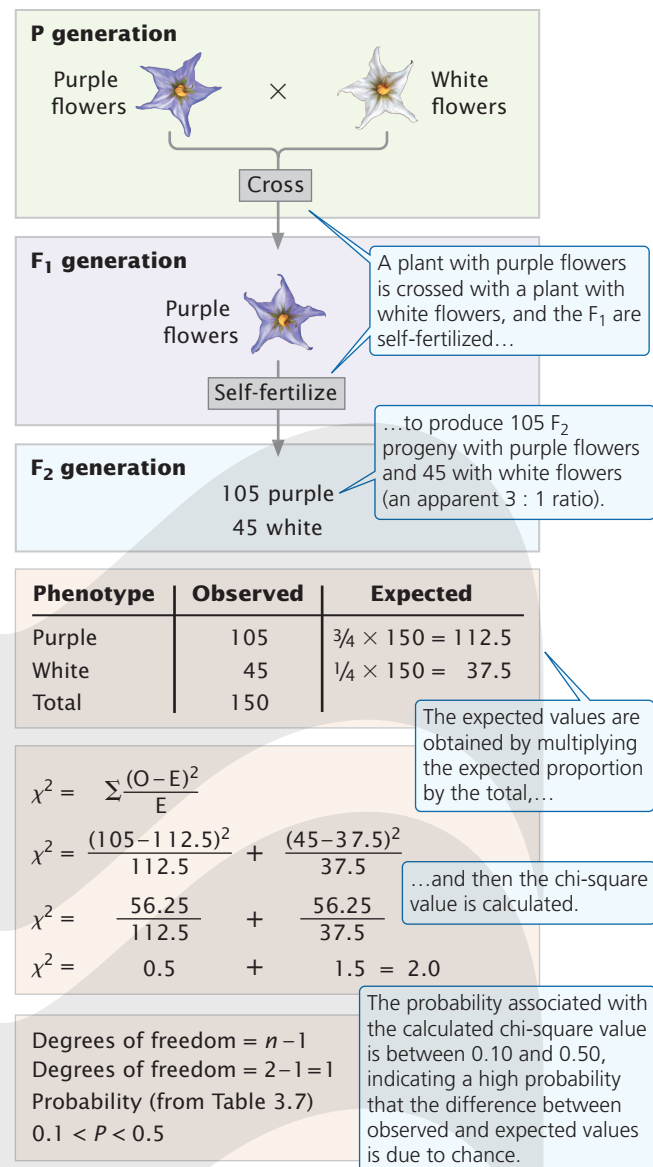
THINK-PAIR-SHARE Question 8

## CONCEPTS

Differences between observed and expected ratios among the progeny of a cross can arise by chance. The chi-square goodness-of-fit test can be used to evaluate whether deviations between observed and expected numbers 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



| Phenotype | Observed | Expected |
|---|---|---|
| Purple | 105 | $\frac{3}{4} \times 150 = 112.5$ |
| White | 45 | $\frac{1}{4} \times 150 = 37.5$ |
| Total | 150 | |

The expected values are obtained by multiplying the expected proportion by the total,…

$$\chi^2 = \Sigma \frac{(O - E)^2}{E}$$

$$\chi^2 = \frac{(105 - 112.5)^2}{112.5} + \frac{(45 - 37.5)^2}{37.5}$$

…and then the chi-square value is calculated.

$$\chi^2 = \frac{56.25}{112.5} + \frac{56.25}{37.5}$$

$$\chi^2 = 0.5 + 1.5 = 2.0$$

Degrees of freedom = $n - 1$
Degrees of freedom = $2 - 1 = 1$
Probability (from Table 3.7)
$0.1 < P < 0.5$

The probability associated with the calculated chi-square value is between 0.10 and 0.50, indicating a high probability that the difference between observed and expected values is due to chance.

**Conclusion:** No significant difference between observed and expected values.

**3.15 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.**

## 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.

■ The binomial expansion can be used to determine the probability of a particular combination of 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.

## IMPORTANT TERMS

gene (p. 50)
allele (p. 50)
locus (p. 50)
genotype (p. 51)
homozygous (p. 51)
heterozygous (p. 51)
phenotype (p. 51)
monohybrid cross (p. 51)
P (parental) generation (p. 51)

$F_1$ (first filial) generation (p. 51)
reciprocal cross (p. 51)
$F_2$ (second filial) generation (p. 52)
dominant (p. 52)
recessive (p. 52)
principle of segregation (Mendel's first law) (p. 53)

concept of dominance (p. 53)
chromosome theory of heredity (p. 54)
backcross (p. 56)
Punnett square (p. 56)
probability (p. 57)
multiplication rule (p. 57)
addition rule (p. 57)

conditional probability (p. 58)
testcross (p. 60)
wild type (p. 60)
dihybrid cross (p. 61)
principle of independent assortment (Mendel's second law) (p. 62)
chi-square goodness-of-fit test (p. 67)

## 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

---

## WORKED PROBLEMS

### Problem 1

Short hair (*S*) in rabbits is dominant to long hair (*s*). The following crosses are carried out, producing the progeny shown. Give all possible genotypes of the parents in each cross.

| | Parents | Progeny |
|---|---|---|
| **a.** | short × short | 4 short and 2 long |
| **b.** | short × short | 8 short |
| **c.** | short × long | 12 short |
| **d.** | short × long | 3 short and 1 long |
| **e.** | long × long | 2 long |

#### ›› Solution Strategy

**What information is required in your answer to the problem?**

All possible genotypes of the parents in each cross.

**What information is provided to solve the problem?**

- Short hair is dominant to long hair.
- Phenotypes of the parents in each cross.
- Phenotypes and number of progeny of each cross.

**Note:** The problem asks for all possible genotypes of the parents.

**For help with this problem, review:**
Connecting Concepts: Ratios in Simple Crosses in Section 3.2.

#### ›› Solution Steps

For this problem, it is useful to first gather as much information about the genotypes of the parents as possible on the basis of their phenotypes. We can then look at the types of progeny produced to provide the missing information.

**a.** short × short        4 short and 2 long

Because short hair is dominant to long hair, a rabbit with short hair could be either *SS* or *Ss*. The two long-haired offspring must be homozygous (*ss*) because long hair is recessive and will appear in the phenotype only when two alleles for long hair are present. Because each parent contributes one of the two alleles found in the progeny, each parent must be carrying the *s* allele and must therefore be *Ss*.

**Note:** When both parents are heterozygotes (*Ss* × *Ss*), we expect to see a 3 : 1 ratio in progeny, but just by chance the progeny exhibited a 4 : 2 ratio.

**b.** short × short        8 short

The short-haired parents could be *SS* or *Ss*. All eight of the offspring are short haired (*S_*), so at least one of the parents is likely to be homozygous (*SS*); if both parents were heterozygous, we would expect $\frac{1}{4}$ of the progeny to be long haired (*ss*), but we do not observe any long-haired progeny. The other parent could be homozygous (*SS*) or heterozygous (*Ss*); as long as one parent is homozygous, all the offspring will be short haired. It is theoretically possible, although unlikely, that both parents are heterozygous (*Ss* × *Ss*). If both were heterozygous, we would expect two of the eight progeny to be long haired. Although no long-haired progeny are observed, it is possible that just by chance no long-haired rabbits were produced among the eight progeny of the cross.

**c.** short × long        12 short

The short-haired parent could be *SS* or *Ss*. The long-haired parent must be *ss*. If the short-haired parent were heterozygous (*Ss*), half of the offspring would be expected to be long haired, but we don't see any long-haired progeny. Therefore, this parent is most likely homozygous (*SS*). It is theoretically possible, although unlikely, that the parent is heterozygous and just by chance no long-haired progeny were produced.

**d.** short × long        3 short and 1 long

On the basis of its phenotype, the short-haired parent could be homozygous (*SS*) or heterozygous (*Ss*), but the presence of one long-haired offspring tells us that the short-haired parent must be heterozygous (*Ss*). The long-haired parent must be homozygous (*ss*).

**e.** long × long        2 long

Because long hair is recessive, both parents must be homozygous for a long-hair allele (*ss*).

## Problem 2

In cats, black coat color is dominant to gray. A female black cat whose mother is gray mates with a gray male. If this female has a litter of six kittens, what is the probability that three will be black and three will be gray?

### ›› Solution Strategy

**What information is required in your answer to the problem?**
The probability that in a litter of six kittens, three will be black and three will be gray.

**What information is provided to solve the problem?**

- Black is dominant to gray.
- The mother of the litter is black and her mother is gray.
- The father of the litter is gray.

**For help with this problem, review:**
The Binomial Expansion and Probability in Section 3.2.

### ›› Solution Steps

**Hint:** We can determine the female parent's genotype from her phenotype and her mother's phenotype.

Because black ($G$) is dominant to gray ($g$), a black cat may be homozygous ($GG$) or heterozygous ($Gg$). The black female in this problem must be heterozygous ($Gg$) because her mother is gray ($gg$) and she must have inherited one of her mother's alleles. The gray male must be homozygous ($gg$) because gray is recessive. Thus, the cross is

$$Gg \qquad \times \qquad gg$$
Black female          Gray male

$$\downarrow$$

$\frac{1}{2}\ Gg$ black
$\frac{1}{2}\ gg$ gray
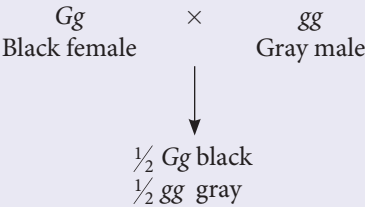
**Recall:** The binomial expansion can be used to determine the probability of different combinations of traits in the progeny of a cross.

We can use the binomial expansion to determine the probability of obtaining three black and three gray kittens in a litter of six. Let $p$ equal the probability of a kitten being black and $q$ equal the probability of a kitten being gray. The binomial is $(p+q)^6$, the expansion of which is

**Hint:** See pp. 58–60 for an explanation of how to expand the binomial.

$$(p+q)^6 = p^6 + 6p^5q^1 + 15p^4q^2 + 20p^3q^3 + 15p^2q^4 + 6p^1q^5 + q^6$$

The probability of obtaining three black and three gray kittens in a litter of six is provided by the term $20p^3q^3$. The probabilities of $p$ and $q$ are both $\frac{1}{2}$, so the overall probability is $20(\frac{1}{2})^3(\frac{1}{2})^3 = \frac{20}{64} = \frac{5}{16}$.

## Problem 3

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

**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.

In this cross, purple × yellow produces approximately $\frac{1}{2}$ purple and $\frac{1}{2}$ 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 ($pp$).

Now let's examine the other character. Full × shrunken produces $\frac{1}{2}$ full and $\frac{1}{2}$ 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* $= \frac{1}{2}$ purple × $\frac{1}{2}$ full $= \frac{1}{4}$ purple, full

     *Pp ff* $= \frac{1}{2}$ purple × $\frac{1}{2}$ shrunken $= \frac{1}{4}$ purple, shrunken

     *Pp Ff* $= \frac{1}{2}$ yellow × $\frac{1}{2}$ full $= \frac{1}{4}$ yellow, full

     *Pp ff* $= \frac{1}{2}$ yellow × $\frac{1}{2}$ shrunken $= \frac{1}{4}$ yellow, shrunken

Our genetic calculations predict that from this cross, we should see $\frac{1}{4}$ purple, full-kernel progeny; $\frac{1}{4}$ purple, shrunken-kernel progeny; $\frac{1}{4}$ yellow, full-kernel progeny; and $\frac{1}{4}$ 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 | $\frac{1}{4} \times 400 = 100$ |

**Hint:** See Figure 3.15 for help on how to carry out a chi-square test.

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

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

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

$$+ \frac{(91 - 100)^2}{100} + \frac{(94 - 100)^2}{100}$$

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

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

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

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 3.7). 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). The probability ($P$) associated with the calculated chi-square value is therefore $0.5 > P > 0.1$. 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.

## 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?

## APPLICATION QUESTIONS AND PROBLEMS

### Introduction

12. 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

*13. 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

*14. 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.

15. **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?

16. 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, 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.

*17. 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).



[Wegner/ARCO/Nature Picture Library; 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?

*18. 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 |

19. **Figure 3.8** 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?

20. 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.

*21. In sheep, lustrous fleece results from an allele ($L$) that is dominant to an allele ($l$) for normal fleece. A ewe (adult female) with lustrous fleece is mated with a ram (adult male) with normal fleece. The ewe then gives birth to a single lamb with normal fleece. From this single offspring, is it possible to determine the genotypes of the two parents? If so, what are their genotypes? If not, why not?

[Jeffrey van Daele/FeaturePics.]

*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. In rabbits, coat color is a genetically determined trait. Some black females always produce black progeny, whereas other black females produce black progeny and white progeny. Explain how this occurs.

24. 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?

*25. 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

*26. 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

*27. In a family of seven children, what is the probability of obtaining the following distribution of boys and girls?

   a. All boys

   b. All children of the same sex

   c. Six girls and one boy

   d. Four boys and three girls

   e. Four girls and three boys

28. 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?

*29. 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

CONNECT

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.

*30. In guinea pigs, the allele for black fur ($B$) is dominant to
CONNECT the allele for brown ($b$) fur. 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.

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

31. 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?

32. **Figure 3.11** 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?

*33. 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

[Jean-Michel Labat/Science Source.]

gray cat homozygous for 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?

*34. The following two genotypes are crossed: *Aa Bb Cc dd Ee* × *Aa bb Cc Dd Ee*. What will the proportion of the following genotypes be 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*

35. In mice, an allele for apricot eyes ($a$) is recessive to an allele for brown eyes ($a^+$). At an independently assorting locus, an allele for tan coat color ($t$) is recessive to an allele for black coat color ($t^+$). A mouse that is homozygous for brown eyes and black coat color is crossed with a mouse having apricot eyes and a tan coat. The resulting F$_1$ are intercrossed to produce the F$_2$. In a litter of eight F$_2$ mice, what is the probability that two will have apricot eyes and tan coats?

36. 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.

*37. Alleles $A$ and $a$ are located on a pair of metacentric
CONNECT chromosomes. Alleles $B$ and $b$ are located on a pair of acrocentric chromosomes (see 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

*38. J. A. Moore investigated the inheritance of spotting
DATA patterns in leopard frogs (J. A. Moore. 1943. *Journal of
ANALYSIS *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.
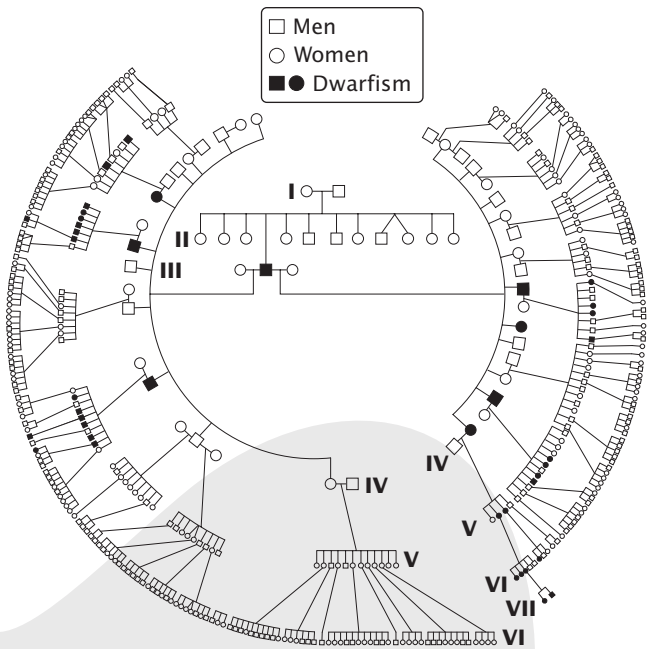
| Parent phenotypes | Progeny phenotypes |
|---|---|
| burnsi × burnsi | 39 burnsi, 6 pipiens |
| burnsi × pipiens | 23 burnsi, 33 pipiens |
| burnsi × pipiens | 196 burnsi, 210 pipiens |

**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.

**39.** In the 1800s, a man with dwarfism who lived in Utah produced a large number of descendants: 22 children, 49 grandchildren, and 250 great-grandchildren (see the illustration of a family pedigree to the right), many of whom also exhibited dwarfism (F. F. Stephens. 1943. *Journal of Heredity* 34:229–235). The type of dwarfism found in this family is called Schmid-type metaphyseal chondrodysplasia, although it was originally thought to be achondroplastic dwarfism. Among the families of this kindred, dwarfism appeared only in members who had one parent with dwarfism. When one parent exhibited dwarfism, the following numbers of children were produced.

| Family in which one parent had dwarfism | Children with normal stature | Children with dwarfism |
|---|---|---|
| A | 15 | 7 |
| B | 4 | 6 |
| C | 1 | 6 |
| D | 6 | 2 |
| E | 2 | 2 |
| F | 8 | 4 |
| G | 4 | 4 |
| H | 2 | 1 |
| I | 0 | 1 |
| J | 3 | 1 |
| K | 2 | 3 |
| L | 2 | 1 |
| M | 2 | 0 |
| N | 1 | 0 |
| O | 0 | 2 |
| Total | 52 | 40 |



[Data from *the Journal of Heredity* 34:232.]

**a.** With the assumption that Schmid-type metaphyseal chondrodysplasia is rare, is this type of dwarfism inherited as a dominant or a recessive trait? Explain your reasoning.

**b.** On the basis of your answer for part *a*, what is the expected ratio of children with dwarfism to children with normal stature in the families given in the table? Use a chi-square test to determine if the total number of children with each phenotype in these families (52 with normal stature, 40 with dwarfism) is significantly different from the number expected.

**c.** Use chi-square tests to determine if the numbers of children with each phenotype in family C (1 with normal stature, 6 with dwarfism) and in family D (6 with normal stature, 2 with dwarfism) are significantly different from the numbers expected on the basis of your proposed mode of inheritance. How would you explain these deviations from the overall ratio expected?

**40.** Pink-eye and albino are two recessive traits found in the deer mouse *Peromyscus maniculatus*. In pink-eyed mice, the eye is devoid of color and appears pink because of the blood vessels within it. Albino mice are completely lacking color, both in their fur and in their eyes. F. H. Clark crossed pink-eyed mice with albino mice; the resulting $F_1$ had normal coloration in their fur and eyes. He then crossed these $F_1$ mice with mice that were pink eyed and albino and obtained the following progeny. It is hard to distinguish between mice that are albino and mice that are both pink eyed and albino, so he combined these two phenotypes (F. H. Clark. 1936. *Journal of Heredity* 27:259–260).

| Phenotype | Number of progeny |
|---|---|
| wild-type fur, wild-type eye color | 12 |
| wild-type fur, pink-eye | 62 |
| albino<br>albino, pink-eye | 78 |
| Total | 152 |

a. Give the expected numbers of progeny with each phenotype if the genes for pink-eye and albino assort independently.

b. Use a chi-square test to determine if the observed numbers of progeny fit the number expected with independent assortment.

*41. 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.

## CHALLENGE QUESTIONS

### Section 3.2

42. Dwarfism is a recessive trait in Hereford cattle; the gene causing this trait has not been identified. A rancher in western Texas discovers that several of the calves in his herd are dwarfs, and he wants to eliminate this undesirable trait from the herd as rapidly as possible. Suppose that the rancher hires you as a genetic consultant to advise him on how to breed the dwarfism trait out of the herd. What crosses would you advise the rancher to conduct to ensure that the allele causing dwarfism is eliminated from the herd?

*43. 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.

44. 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 $\frac{1}{4}$ of their children. To his surprise, the geneticist finds that the frequency of albinism among the children of these families is significantly greater than $\frac{1}{4}$. Can you think of an explanation for the higher-than-expected frequency of albinism among these families?

45. Two distinct phenotypes are found in the salamander *Plethodon cinereus*: a red form and a black form. Some biologists have speculated that the red phenotype is due to an allele that is dominant to an allele for black. Unfortunately, these salamanders will not mate in captivity, so the hypothesis that red is dominant to black has never been tested.

One day, a genetics student is hiking through the forest and finds 30 female salamanders, some red and some black, laying eggs. The student places each female with her eggs (about 20 to 30 eggs per female) in a separate plastic bag and takes them back to the lab. There, the student successfully raises the eggs until they hatch. After the eggs have hatched, the student records the phenotypes of the juvenile salamanders, along with the phenotypes of their mothers. Thus, the student has the phenotypes for 30 females and their progeny, but no information is available about the phenotypes of the fathers.

[George Grall/Getty Images.]

Explain how the student can determine whether red is dominant to black with this information on the phenotypes of the females and their offspring.

## 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 genes for blond hair?

### Section 3.1

2. 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

3. Geneticists often carry out reciprocal crosses when they are studying the inheritance of traits. Why do geneticists use reciprocal crosses?

4. 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 ¾ 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 ¾ red-haired children. Who is correct and why?

5. Suppose that you are raising Mongolian gerbils. You notice that some of your gerbils have white spots, whereas others have solid coats. What type of crosses could you carry out to determine whether white spots are due to a recessive or a dominant allele?

### Section 3.3

6. 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?

7. In cats, short hair ($L$) is dominant to long hair ($l$), and stripes ($A$) are dominant to solid color ($a$). A cat with genotype $Ll\ Aa$ mates with a cat that is $Ll\ aa$, and they produce a litter of six kittens. What is the probability that four of the six kittens will have both long hair and stripes?

### 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 and starchy kernel is crossed with a plant grown from a yellow and 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

### Introduction

1. Draw a concept map that explains the genetic basis of blond hair in Solomon Islanders using the following terms: Solomon Islanders, Europeans, blond hair, recessive trait, dark hair, melanin, chromosome 9, dominant trait, *TYRP1*, *KITLG*, *TT*, *CT*, *CC*.

### Section 3.1

2. 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

3. 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).