

# 6.1

## Chromosomes and Meiosis

**KEY CONCEPT** Gametes have half the number of chromosomes that body cells have.

### ▶ MAIN IDEAS

- You have body cells and gametes.
- Your cells have autosomes and sex chromosomes.
- Body cells are diploid; gametes are haploid.

### VOCABULARY

**somatic cell**, p. 168

**gamete**, p. 168

**homologous chromosome**, p. 169

**autosome**, p. 169

**sex chromosome**, p. 169

**sexual reproduction**, p. 170

**fertilization**, p. 170

**diploid**, p. 170

**haploid**, p. 170

**meiosis**, p. 170



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**Connect** Perhaps you are familiar with the saying, “Everything old is new again.” This phrase usually indicates that a past style is again current. However, it applies equally well to you. The fusion of a single egg and sperm cell resulted in the complex creature that is you. There’s never been anyone quite like you. And yet the DNA that directs your cells came from your mother and father. And their DNA came from their mother and father, and so on and so on. In this chapter, you will examine the processes that went into making you who you are.

### ▶ MAIN IDEA

## You have body cells and gametes.

You have many types of specialized cells in your body, but they can be divided into two major groups: somatic cells and germ cells. **Somatic cells** (soh-MAT-ihk), also called body cells, make up most of your body tissues and organs. For example, your spleen, kidneys, and eyeballs are all made entirely of body cells. DNA in your body cells is not passed on to your children. Germ cells, in contrast, are cells in your reproductive organs, the ovaries or the testes, that develop into gametes. **Gametes** are sex cells—ova, or eggs, in the female, and spermatozoa, or sperm cells, in the male. DNA in your gametes can be passed on to your children.

Each species has a characteristic number of chromosomes per cell. This number is typically given for body cells, not for gametes. Chromosome number does not seem to be related to the complexity of an organism. For example, yeast have 32 chromosomes, which come in 16 pairs. The fruit flies commonly used in genetic experiments have 8 chromosomes, which come in 4 pairs. A fern holds the record for the most chromosomes—more than 1200. Each of your body cells contains a set of 46 chromosomes, which come in 23 pairs. These cells are genetically identical to each other unless mutations have occurred. As you learned in Chapter 5, cells within an organism differ from each other because different genes are expressed, not because they have different genes.

### TAKING NOTES

Make a two-column table to keep track of the vocabulary in this chapter.

Term	Definition
somatic cell	
gamete	

**Identify** Which cell type makes up the brain?

**MAIN IDEA**

## Your cells have autosomes and sex chromosomes.

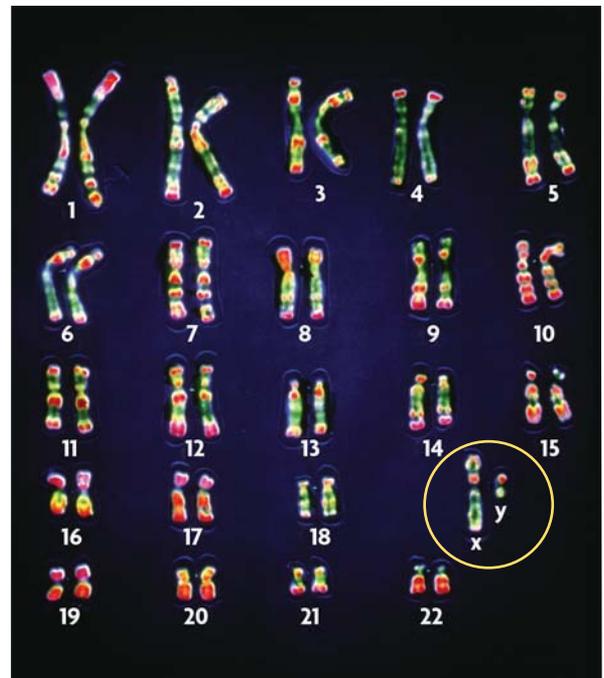
Suppose you had 23 pairs of gloves. You would have a total of 46 gloves that you could divide into two sets, 23 right and 23 left. Similarly, your body cells have 23 pairs of chromosomes for a total of 46 that can be divided into two sets: 23 from your mother and 23 from your father. Just as you use both gloves when it's cold outside, your cells use both sets of chromosomes to function properly.

Together, each pair of chromosomes is referred to as a homologous pair. In this context, *homologous* means “having the same structure.” **Homologous chromosomes** are two chromosomes—one inherited from the mother, one from the father—that have the same length and general appearance. More importantly, these chromosomes have copies of the same genes, although the two copies may differ. For example, if you have a gene that influences blood cholesterol levels on chromosome 8, you will have one copy from your mother and one copy from your father. It is possible that one of these copies is associated with high cholesterol levels, while the other is associated with low cholesterol levels. For convenience, scientists have assigned a number to each pair of homologous chromosomes, ordered from largest to smallest. As **FIGURE 6.1** shows, the largest pair of chromosomes is number 1, the next largest pair is number 2, and so forth.

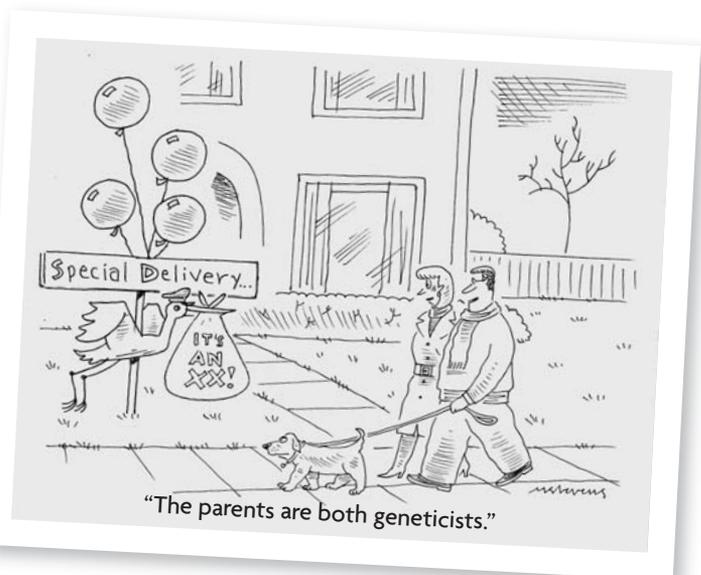
Collectively, chromosome pairs 1 through 22 make up your **autosomes**, chromosomes that contain genes for characteristics not directly related to the sex of an organism. But what about the 23rd chromosome pair?

Most sexually reproducing species also have **sex chromosomes** that directly control the development of sexual characteristics. Humans have two very different sex chromosomes, X and Y. How sex is determined varies by species. In all mammals, including humans, an organism's sex is determined by the XY system. An organism with two X chromosomes is female. An organism with one X and one Y chromosome is male. Sex chromosomes make up your 23rd pair of chromosomes. Although the X and Y chromosomes pair with each other, they are not homologous. The X chromosome is the larger sex chromosome and contains numerous genes, including many that are unrelated to sexual characteristics. The Y chromosome is the sex chromosome that contains genes that direct the development of the testes and other male traits. It is the smallest chromosome and carries the fewest genes.

**Summarize** Are homologous chromosomes identical to each other? Explain.



**FIGURE 6.1** Human DNA is organized into two sets of 23 chromosomes. Each set contains 22 autosomes and 1 sex chromosome. Females have two X chromosomes. Males have an X and a Y chromosome (circled). (colored LM; magnification 4400 $\times$ )



**MAIN IDEA**

## Body cells are diploid; gametes are haploid.

**Sexual reproduction** involves the fusion of two gametes that results in offspring that are a genetic mixture of both parents. The actual fusion of an egg and a sperm cell is called **fertilization**. When fertilization occurs, the nuclei of the egg and sperm cell fuse to form one nucleus. This new nucleus must have the correct number of chromosomes for a healthy new organism to develop. Therefore, the egg and sperm cell need only half the usual number of chromosomes—one chromosome from each homologous pair.

### Diploid and Haploid Cells

Body cells and gametes have different numbers of chromosomes. Your body cells are diploid. **Diploid** (DIHP-LOYD) means a cell has two copies of each chromosome: one copy from the mother, and one copy from the father. Diploid cells can be represented as  $2n$ . In humans, the diploid chromosome number is 46.

Gametes are not diploid cells; they are haploid cells, represented as  $n$ .

**Haploid** (HAP-LOYD) means that a cell has only one copy of each chromosome. Each human egg or sperm cell has 22 autosomes and 1 sex chromosome. In the egg, the sex chromosome is always an X chromosome. In the sperm cell, the sex chromosome can be an X chromosome or a Y chromosome. The reason for this difference will be discussed in the following sections.

Maintaining the correct number of chromosomes is important to the survival of all organisms. Typically, a change in chromosome number is harmful. However, increasing the number of sets of chromosomes can, on occasion, give rise to a new species. The fertilization of nonhaploid gametes has played an important role in plant evolution by rapidly making new species with more than two sets of chromosomes. For example, some plants have four copies of each chromosome, a condition called tetraploidy ( $4n$ ). This type of event has occurred in many groups of plants, but it is very rare in animals.

### Meiosis

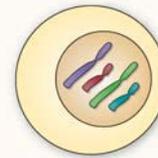
Germ cells in your reproductive organs undergo the process of meiosis to form gametes. **Meiosis** (my-OH-sihs) is a form of nuclear division that divides a diploid cell into haploid cells. This process is essential for sexual reproduction. The details of meiosis will be presented in the next section. But **FIGURE 6.2** highlights some differences between mitosis and meiosis in advance to help you keep these two processes clear in your mind.

#### VOCABULARY

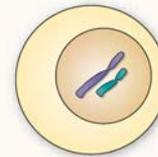
*Diploid* comes from the Greek word *diplous*, which means “double.” *Haploid* comes from the Greek word *haplous*, which means “single.”

#### VISUAL VOCAB

**Diploid** cells have two copies of each chromosome: one copy from the mother and one from the father.



Body cells are diploid ( $2n$ ).



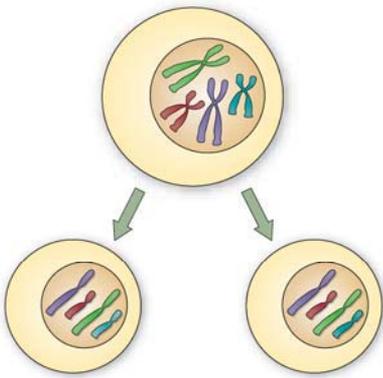
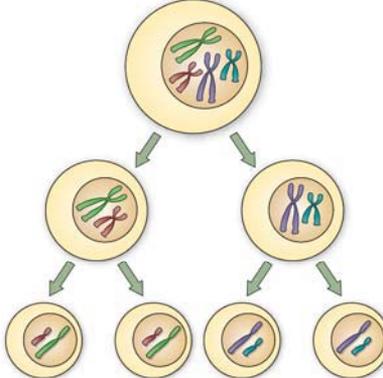
Gametes (sex cells) are haploid ( $n$ ).

**Haploid** cells have only one copy of each chromosome.

#### Connecting CONCEPTS

**Plant Life Cycles** As you will learn in **Chapter 22**, all plants complete their life cycle by alternating between two phases: diploid and haploid. During the diploid phase, plants make spores. During the haploid phase, plants make gametes.

**FIGURE 6.2 Comparing Mitosis and Meiosis**

MITOSIS		MEIOSIS	
	Produces genetically identical cells		Produces genetically unique cells
	Results in diploid cells		Results in haploid cells
	Takes place throughout an organism's lifetime		Takes place only at certain times in an organism's life cycle
	Involved in asexual reproduction		Involved in sexual reproduction

**Compare** Using the diagrams above, explain how you think the process of meiosis differs from mitosis.

In Chapter 5 you learned about mitosis, another form of nuclear division. Recall that mitosis is a process that occurs in body cells. It helps produce daughter cells that are genetically identical to the parent cell. In cells undergoing mitosis, DNA is copied once and divided once. Both the parent cell and the daughter cells are diploid. Mitosis is used for development, growth, and repair in all types of organisms. It is also used for reproduction in asexually reproducing eukaryotes.

In contrast, meiosis occurs in germ cells to produce gametes. This process is sometimes called a “reduction division” because it reduces chromosome number by half. In cells undergoing meiosis, DNA is copied once but divided twice. Meiosis makes genetically unique haploid cells from a diploid cell. These haploid cells then undergo more processing in the ovaries or testes, finally forming mature gametes.

**Apply** Why is it important that gametes are haploid cells?

## 6.1 ASSESSMENT



### REVIEWING MAIN IDEAS

- Where are germ cells located in the human body?
- What is the difference between an **autosome** and a **sex chromosome**?
- Is the cell that results from **fertilization** a **haploid** or **diploid** cell? Explain.

### CRITICAL THINKING

- Infer** Does mitosis or **meiosis** occur more frequently in your body? Explain your answer.
- Analyze** Do you think the Y chromosome contains genes that are critical for an organism's survival? Explain your reasoning.

### Connecting CONCEPTS

- Telomeres** The ends of DNA molecules form telomeres that help keep the ends of chromosomes from sticking to each other. Why might this be especially important in germ cells, which go through meiosis and make haploid **gametes**?

# 6.2

## Process of Meiosis

**KEY CONCEPT** During meiosis, diploid cells undergo two cell divisions that result in haploid cells.

### ▶ MAIN IDEAS

- Cells go through two rounds of division in meiosis.
- Haploid cells develop into mature gametes.

### VOCABULARY

**gametogenesis**, p. 176

**sperm**, p. 176

**egg**, p. 176

**polar body**, p. 176



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**Connect** Sometimes division is difficult, such as splitting the bill at a restaurant or dividing people into teams for basketball. Luckily, understanding how meiosis divides chromosomes between cells is not that hard. Meiosis begins with a diploid cell that has already undergone DNA replication. The cell copies the chromosomes once and divides them twice, making four haploid cells.

### ▶ MAIN IDEA

## Cells go through two rounds of division in meiosis.

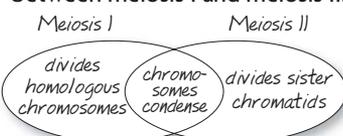
Meiosis is a form of nuclear division that creates four haploid cells from one diploid cell. This process involves two rounds of cell division—meiosis I and meiosis II. Each round of cell division has four phases, which are similar to those in mitosis. To keep the two processes distinct in your mind, focus on the big picture. Pay attention to how meiosis reduces chromosome number and creates genetic diversity.

### Connecting CONCEPTS

**Mitosis** As you learned in Chapter 5, a condensed, duplicated chromosome is made of two chromatids. Sister chromatids separate during anaphase in mitosis.

### TAKING NOTES

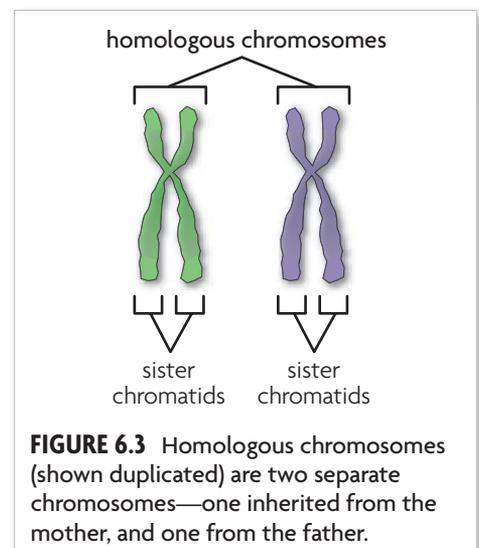
Draw a Venn diagram like the one below to summarize the similarities and differences between meiosis I and meiosis II.



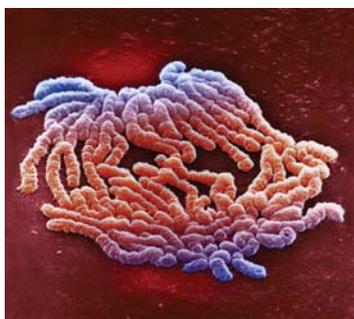
### Homologous Chromosomes and Sister Chromatids

To understand meiosis, you need to distinguish between homologous chromosomes and sister chromatids. As **FIGURE 6.3** shows, homologous chromosomes are two separate chromosomes: one from your mother, one from your father. Homologous chromosomes are very similar to each other, since they have the same length and carry the same genes. But they are not copies of each other. In contrast, each half of a duplicated chromosome is called a chromatid. Together, the two chromatids are called sister chromatids. Thus, *sister chromatids* refers to the duplicated chromosomes that remain attached (by the centromere).

Homologous chromosomes are divided in meiosis I. Sister chromatids are not divided until meiosis II.



**FIGURE 6.3** Homologous chromosomes (shown duplicated) are two separate chromosomes—one inherited from the mother, and one from the father.



**FIGURE 6.4** Homologous chromosomes separate during anaphase I. (colored SEM; magnification 2200×)

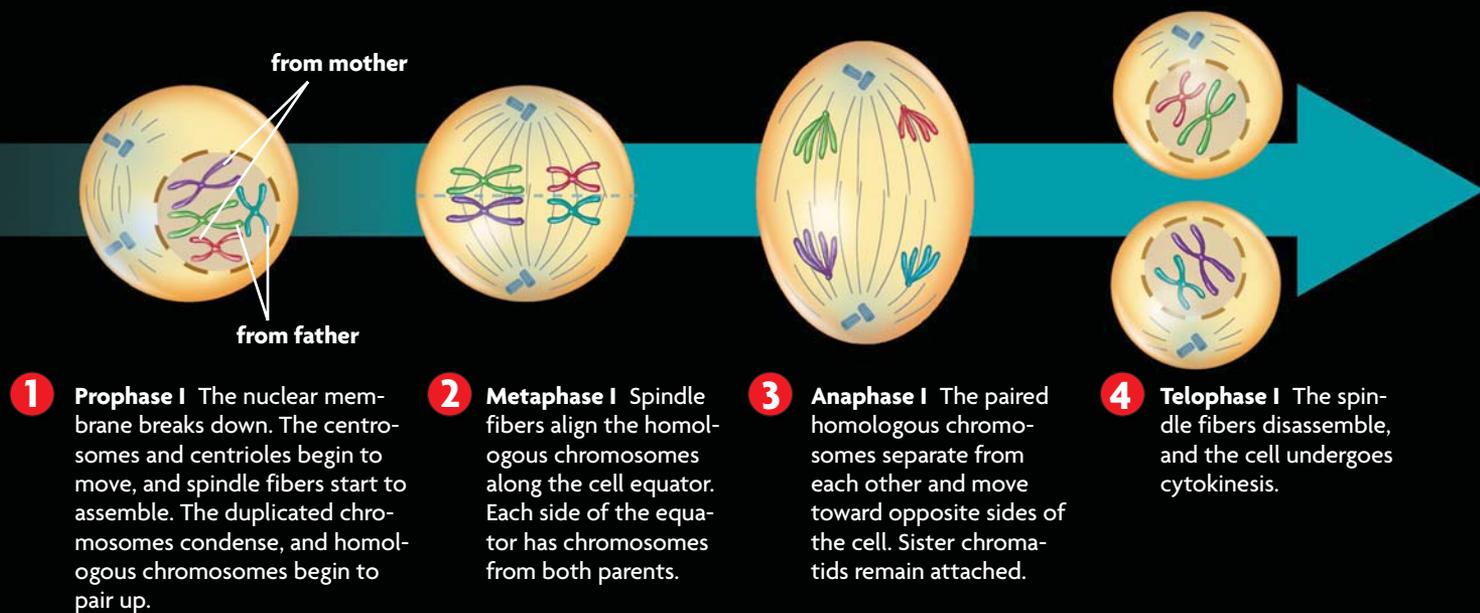
## Meiosis I

Before meiosis begins, DNA has already been copied. Meiosis I divides homologous chromosomes, producing two haploid cells with duplicated chromosomes. Like mitosis, scientists describe meiosis in terms of phases, illustrated in **FIGURE 6.5** below. The figure is simplified, showing only four chromosomes.

- 1 Prophase I** Early in meiosis, the nuclear membrane breaks down, the centrosomes and centrioles move to opposite sides of the cell, and spindle fibers start to assemble. The duplicated chromosomes condense, and homologous chromosomes pair up. They appear to pair up precisely, gene for gene, down their entire length. The sex chromosomes also pair with each other, and some regions of their DNA appear to line up as well.
- 2 Metaphase I** The homologous chromosome pairs are randomly lined up along the middle of the cell by spindle fibers. The result is that 23 chromosomes—some from the father, some from the mother—are lined up along each side of the cell equator. This arrangement mixes up the chromosomal combinations and helps create and maintain genetic diversity. Since human cells have 23 pairs of chromosomes, meiosis may result in  $2^{23}$ , or 8,388,608, possible combinations of chromosomes.
- 3 Anaphase I** Next, the paired homologous chromosomes separate from each other and move toward opposite sides of the cell. The sister chromatids remain together during this step and throughout meiosis I.
- 4 Telophase I** The nuclear membrane forms again in some species, the spindle fibers disassemble, and the cell undergoes cytokinesis. The end result is two cells that each have a unique combination of 23 duplicated chromosomes coming from both parents.

## FIGURE 6.5 Meiosis

Meiosis I divides homologous chromosomes.



## Meiosis II

Meiosis II divides sister chromatids, and results in undoubled chromosomes. The following description of this process applies to both of the cells produced in meiosis I. Note that DNA is not copied again between these two stages.

- 5 Prophase II** The nuclear membrane breaks down, centrosomes and centrioles move to opposite sides of the cell, and spindle fibers assemble.
- 6 Metaphase II** Spindle fibers align the 23 chromosomes at the cell equator. Each chromosome still has two sister chromatids at this stage.
- 7 Anaphase II** Next, the sister chromatids are pulled apart from each other and move to opposite sides of the cell.
- 8 Telophase II** Finally, nuclear membranes form around each set of chromosomes at opposite ends of the cell, the spindle fibers break apart, and the cell undergoes cytokinesis. The end result is four haploid cells with a combination of chromosomes from both the mother and father.

Now that you've seen how meiosis works, let's review some key differences between the processes of meiosis and mitosis.

- Meiosis has two cell divisions. Mitosis has only one cell division.
- During meiosis, homologous chromosomes pair up along the cell equator. During mitosis, homologous chromosomes never pair up.
- In anaphase I of meiosis, sister chromatids remain together. In anaphase of mitosis, sister chromatids separate.
- Meiosis results in haploid cells. Mitosis results in diploid cells.

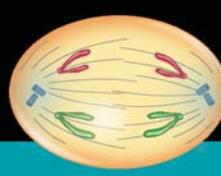
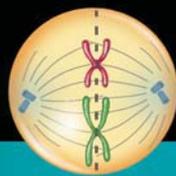
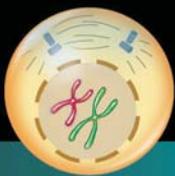
**Contrast** What is the major difference between metaphase I and metaphase II?

### Connecting CONCEPTS

**Cytokinesis** As you learned in Chapter 5, cytokinesis is the division of the cell cytoplasm. This process is the same in cells undergoing either mitosis or meiosis.



Meiosis II divides sister chromatids. The overall process produces haploid cells.



- 5 Prophase II** The centrosomes and centrioles move to opposite sides of the cell, and spindle fibers start to assemble.

- 6 Metaphase II** Spindle fibers align the chromosomes along the cell equator.

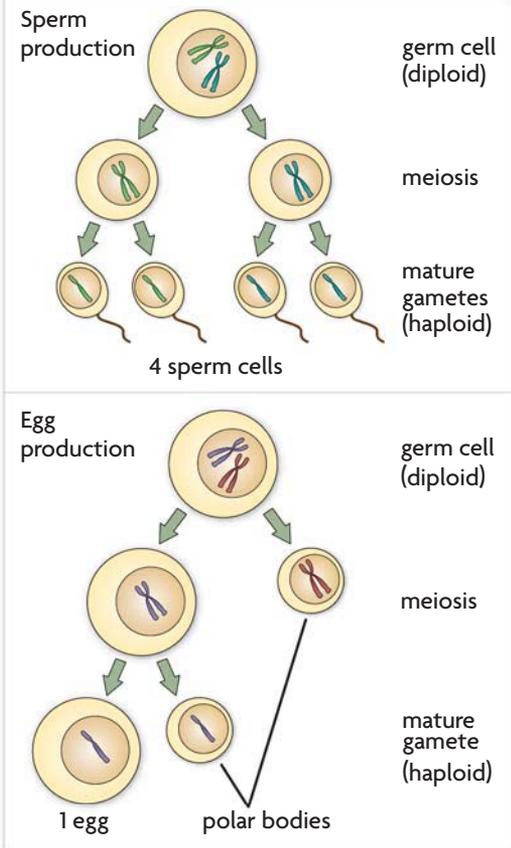
- 7 Anaphase II** The sister chromatids are pulled apart from each other and move to opposite sides of the cell.

- 8 Telophase II** The nuclear membranes form again around the chromosomes, the spindle fibers break apart, and the cell undergoes cytokinesis.

**▶ MAIN IDEA**

## Haploid cells develop into mature gametes.

**FIGURE 6.6 GAMETOGENESIS**



Haploid cells are the end result of meiosis. Yet these cells are incapable of fertilization until they go through more changes to form mature gametes. **Gametogenesis** (guh-MEE-tuh-JEHN-ih-sihs) is the production of gametes. As **FIGURE 6.6** shows, gametogenesis includes both meiosis and other changes that produce a mature cell. The final stages of gametogenesis differ between the sexes.

The **sperm** cell, the male gamete, is much smaller than the **egg**, the female gamete. The sperm cell's main contribution to an embryo is DNA. Yet it must swim to an egg to fertilize it, so the ability to move is critical. Sperm formation starts with a round cell and ends by making a streamlined cell that can move rapidly. During this process, significant changes occur. DNA is tightly packed and much of the cytoplasm is lost, forming a compact head. The sperm cell develops a whiplike flagellum and connecting neck region packed with mitochondria that drive the cell. Other changes also take place, such as the addition of new proteins to the cell membrane.

The formation of an egg is a complicated process, as you will read about in greater detail in Chapter 34. It begins before birth, inside the developing body of a female embryo, and is not finished until that egg is fertilized by a sperm many years later. The process includes periods of active development and long periods of inactivity.

An egg not only gives its share of DNA to an embryo but also contributes the organelles, molecular building blocks, and other materials an embryo needs to begin life. Only one of the four cells

produced by each round of meiosis actually makes an egg. One cell—the egg—receives most of the organelles, cytoplasm, and nutrients. Many molecules are not evenly distributed throughout the egg's cytoplasm. This unequal distribution of molecules helps cells in the developing embryo to specialize. The other cells produced by meiosis become **polar bodies**, cells with little more than DNA that are eventually broken down. In many species, including humans, the polar body produced by meiosis I does not undergo meiosis II.

**Apply** Briefly explain how a sperm cell's structure is related to its function.



For more about meiosis, go to [scilinks.org](http://scilinks.org).  
 Keycode: MLB006



## 6.2 ASSESSMENT

### REVIEWING **▶** MAIN IDEAS

- How do homologous chromosomes differ from sister chromatids?
- Explain why an **egg** is so much larger than a **sperm** cell.

### CRITICAL THINKING

- Predict** If, during metaphase I, all 23 maternal chromosomes lined up on one side of the cell, would genetic diversity increase? Explain.
- Contrast** List the key differences between meiosis I and II.

### Connecting CONCEPTS

- Cell Biology** Both mitosis and meiosis are types of nuclear division, but they result in different cell types. Describe how the steps of meiosis I differ from those of mitosis.

# 6.3

## Mendel and Heredity

**KEY CONCEPT** Mendel's research showed that traits are inherited as discrete units.

### ▶ MAIN IDEAS

- Mendel laid the groundwork for genetics.
- Mendel's data revealed patterns of inheritance.

### VOCABULARY

**trait**, p. 177

**genetics**, p. 177

**purebred**, p. 178

**cross**, p. 178

**law of segregation**, p. 179



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**Connect** When a magician makes a coin disappear, you know the coin has not really vanished. You simply cannot see where it is. Maybe it is up a sleeve or in a pocket. When organisms reproduce, some traits seem to disappear too. For centuries, no one could explain why. Then a careful, observant scientist showed that behind this phenomenon were inherited units, or genes.

### ▶ MAIN IDEA

## Mendel laid the groundwork for genetics.

When we think of how offspring resemble or differ from their parents, we typically refer to specific traits. **Traits** are distinguishing characteristics that are inherited, such as eye color, leaf shape, and tail length. Scientists recognized that traits are hereditary, or passed from one generation to the next, long before they understood how traits are passed on. **Genetics** is the study of biological inheritance patterns and variation in organisms.

The groundwork for much of our understanding of genetics was laid in the middle of the 1800s by an Austrian monk named Gregor Mendel, shown in **FIGURE 6.7**. Scientists of the time commonly thought parents' traits were blended in offspring, like mixing red and white paint to get pink paint. But this idea failed to explain how certain traits remained without being "diluted." Mendel, a shrewd mathematician, bred thousands of plants, carefully counting and recording his results. From his data, Mendel correctly predicted the results of meiosis long before chromosomes were discovered. He recognized that traits are inherited as discrete units from the parental generation, like different colored marbles mixed together that can still be picked out separately. By recognizing that organisms inherit two copies of each discrete unit, what we now call genes, Mendel also described how traits were passed between generations.

**Connect** Give two examples of traits not listed above.

### ▶ MAIN IDEA

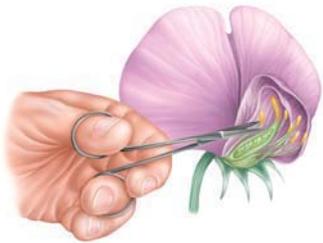
## Mendel's data revealed patterns of inheritance.

Mendel studied plant variation in a monastery garden. He made three key choices about his experiments that played an important role in the development of his laws of inheritance: control over breeding, use of purebred plants, and observation of "either-or" traits that appeared in only two alternate forms.



**FIGURE 6.7** Gregor Mendel is called "the father of genetics" for discovering hereditary units. The significance of his work went unrecognized for almost 40 years.

**FIGURE 6.8 MENDEL'S PROCESS**



Mendel controlled the fertilization of his pea plants by removing the male parts, or stamens.



He then fertilized the female part, or pistil, with pollen from a different pea plant.

### VOCABULARY

In Latin, the word *filius* means “son” and *filia* means “daughter.”

## Experimental Design

Mendel chose pea plants for his experiments because they reproduce quickly, and he could easily control how they mate. The sex organs of a plant are in its flowers, and pea flowers contain both male and female reproductive organs. In nature, the pea flower typically self-pollinates; that is, the plant mates with itself. If a line of plants has self-pollinated for long enough, that line becomes genetically uniform, or **purebred**. As a result, the offspring of purebred parents inherit all of the parent organisms' characteristics. Mendel was able to mate plants with specific traits by interrupting the self-pollination process. As you can see in **FIGURE 6.8**, he removed the male parts of flowers and fertilized the female parts with pollen that contained sperm cells from a different plant. Because he started with purebred plants, Mendel knew that any variations in offspring resulted from his experiments.

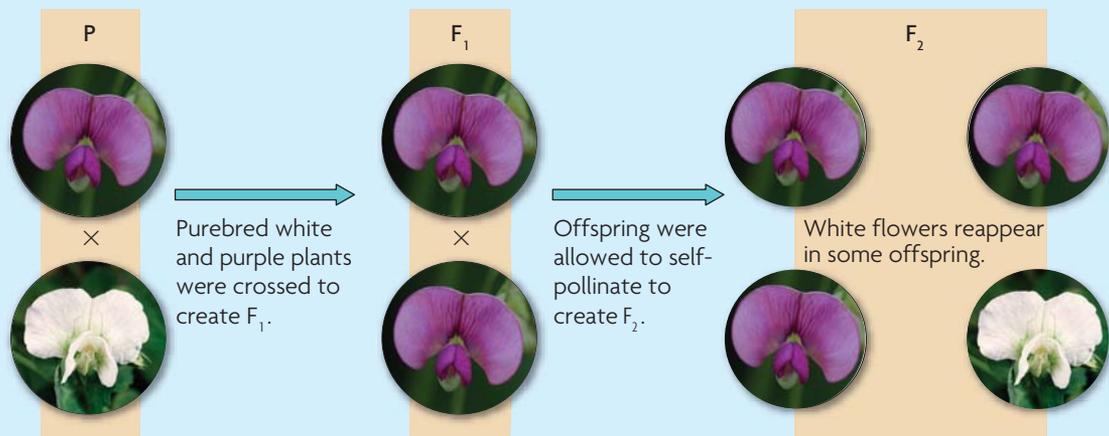
Mendel chose seven traits to follow: pea shape, pea color, pod shape, pod color, plant height, flower color, and flower position. All of these traits are simple “either-or” characteristics; they do not show intermediate features. The plant is tall or short. Its peas are wrinkled or round. What Mendel did not know was that most of the traits he had selected were controlled by genes on separate chromosomes. The selection of these particular traits played a crucial role in enabling Mendel to identify the patterns he observed.

## Results

In genetics, the mating of two organisms is called a **cross**. An example of one of Mendel's crosses is highlighted in **FIGURE 6.9**. In this example, he crossed a purebred white-flowered pea plant with a purebred purple-flowered pea plant. These plants are the parental, or P, generation. The resulting offspring, called the first filial—or  $F_1$ —generation, all had purple flowers. The trait for white flowers seemed to disappear. When Mendel allowed the  $F_1$  generation to self-fertilize, the resulting  $F_2$  generation produced both plants with purple flowers and plants with white flowers. Therefore, the trait for white flowers had not disappeared; it had been hidden, or masked.

**FIGURE 6.9 Mendel's Experimental Cross**

Traits that were hidden when parental purebred flowers were crossed reappeared when the  $F_1$  generation was allowed to self-pollinate.



Mendel did not cross only two plants, however; he crossed many plants. As a result, he was able to observe patterns. He noticed that each cross yielded similar ratios in the  $F_2$  generation: about three-fourths of the plants had purple flowers, and about one-fourth had white flowers. A ratio is a comparison that tells how two or more things relate. This ratio can be expressed as 3:1 (read “three to one”) of purple:white flowers. As you can see in **FIGURE 6.10**, Mendel’s data show this approximately 3:1 ratio for each of his crosses.

FIGURE 6.10 MENDEL'S MONOHYBRID CROSS RESULTS			
$F_2$ TRAITS	DOMINANT	RECESSIVE	RATIO
Pea shape	5474 round	1850 wrinkled	2.96:1
Pea color	6022 yellow	2001 green	3.01:1
Flower color	705 purple	224 white	3.15:1
Pod shape	882 smooth	299 constricted	2.95:1
Pod color	428 green	152 yellow	2.82:1
Flower position	651 axial	207 terminal	3.14:1
Plant height	787 tall	277 short	2.84:1

Source: Mendel, *Abhandlungen* (1865).

## Conclusions

From these observations, Mendel drew three important conclusions. He demonstrated that traits are inherited as discrete units, which explained why individual traits persisted without being blended or diluted over successive generations. Mendel’s two other key conclusions are collectively called the **law of segregation**, or Mendel’s first law.

- Organisms inherit two copies of each gene, one from each parent.
- Organisms donate only one copy of each gene in their gametes. Thus, the two copies of each gene segregate, or separate, during gamete formation.

Section 6.5 covers Mendel’s second law, the law of independent assortment.

**Infer** Explain why Mendel’s choice of either-or characteristics aided his research.

## Connecting CONCEPTS

**Meiosis** As you learned in Section 6.2, during meiosis, homologous chromosomes pair up in prophase I and are separated in anaphase I. The overall process produces haploid cells that have a random assortment of chromosomes.

## 6.3 ASSESSMENT



### REVIEWING MAIN IDEAS

1. Mendel had no understanding of DNA as the genetic material, yet he was able to correctly predict how **traits** were passed between generations. What does Mendel’s work in **genetics** show about the value of scientific observation?
2. Why is it important that Mendel began with **purebred** plants?

### CRITICAL THINKING

3. **Analyze** Mendel saw purple flowers in the  $F_1$  generation, but both purple and white flowers in the  $F_2$ . How did this help him see that traits are inherited as discrete units?
4. **Evaluate** If Mendel had examined only one trait, do you think he would have developed the **law of segregation**? Explain.

## Connecting CONCEPTS

5. **Scientific Process** You have learned that scientific thinking involves observing, forming hypotheses, testing hypotheses, and analyzing data. Use examples from Mendel’s scientific process to show how his work fit this pattern.

# 6.4

## Traits, Genes, and Alleles

**KEY CONCEPT** Genes encode proteins that produce a diverse range of traits.

### ▶ MAIN IDEAS

- The same gene can have many versions.
- Genes influence the development of traits.

### VOCABULARY

**gene**, p. 180

**allele**, p. 180

**homozygous**, p. 180

**heterozygous**, p. 180

**genome**, p. 181

**genotype**, p. 181

**phenotype**, p. 181

**dominant**, p. 181

**recessive**, p. 181



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**Connect** Most things come in many forms. Bread can be wheat, white, or rye. Cars can be two-door, four-door, hatchback, or convertible. Even the variety of potatoes cannot be counted on two hands. Genes, too, come in many forms.

### ▶ MAIN IDEA

## The same gene can have many versions.

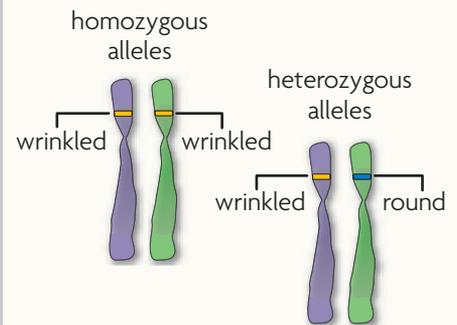
As you have learned, Mendel's discrete units of heredity are now called genes. But what are genes? You can think of a **gene** as a piece of DNA that provides a set of instructions to a cell to make a certain protein. This definition is not precise, but it gives you the main idea. Each gene has a locus, a specific position on a pair of homologous chromosomes. Just as a house is a physical structure and an address tells where that house is located, you can think of the locus as the "address" that tells where a gene is located on a chromosome.

Most genes exist in many forms. In Mendel's experiments, the effects of these different forms were easy to see: yellow or green, round or wrinkled. An **allele** (uh-LEEL) is any of the alternative forms of a gene that may occur at a specific locus. Your cells have two alleles for each gene, one on each of the homologous chromosomes on which the locus for that gene is found. Each parent gives one allele. The two alleles may be the same, or they may be different. The term **homozygous** (HOH-moh-ZY-guhs) describes two of the same alleles at a specific locus. For example, both might code for white flowers. The term **heterozygous** (HEHT-uh-uh-ZY-guhs) describes two different alleles at a specific locus. Thus, one might code for white flowers, the other for purple flowers.

**Compare and Contrast** Distinguish between the terms *locus* and *allele*.

### VISUAL VOCAB

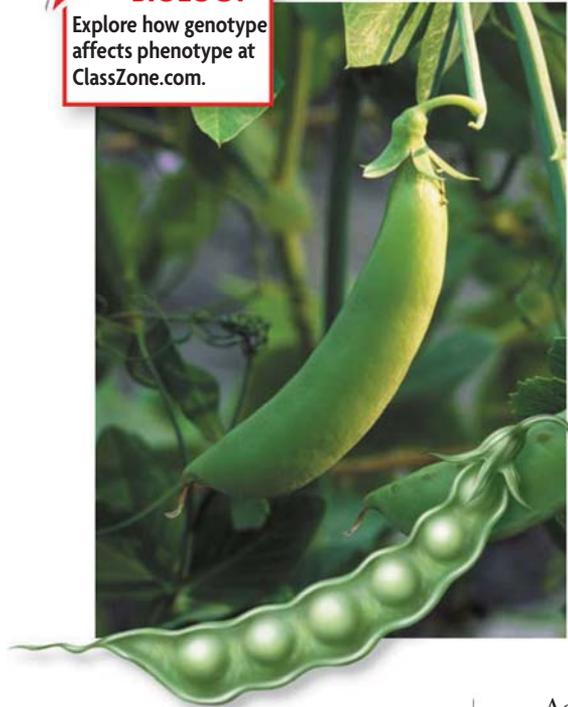
**Homozygous** alleles are identical to each other.



**Heterozygous** alleles are different from each other.

## Animated BIOLOGY

Explore how genotype affects phenotype at ClassZone.com.



**FIGURE 6.12** Both the homozygous dominant and heterozygous genotypes result in smooth, or inflated, pods (top). Only the homozygous recessive genotype results in constricted pods (inset).

A plant's genotype might be homozygous dominant ( $TT$ ), heterozygous ( $Tt$ ), or homozygous recessive ( $tt$ ).

### Alleles and Phenotype

Because some alleles are dominant over others, two genotypes can produce the dominant phenotype. For example, smooth pods and constricted pods in pea plants, shown in **FIGURE 6.12**, are phenotypes. A plant with smooth pods could have a homozygous dominant ( $SS$ ) or heterozygous ( $Ss$ ) genotype. In contrast, a plant with constricted, or compressed, pods could only have a homozygous recessive ( $ss$ ) genotype.

What actually makes one allele dominant over another? The answer is very complicated. It depends on the nature of the protein that is, or is not, made. Let's look at a fairly simple example. Pigment gives cells color. If  $P$  directs flower cells to make pigment, the flower may look purple. If  $p$  directs the cells not to make pigment, the flower looks white. So  $P$  codes for pigment to be present, but  $p$  codes for nothing, the absence of pigment. As a result,  $P$  has to be dominant. Even if the flower has only one  $P$  allele ( $Pp$ ), that one allele tells its cells to make pigment, and the flower has color. Flower pigment is only one example. Many factors make one allele dominant over another.

As you know, most plants are not simply tall or short. Most flowers are not just white or purple. Most traits occur in a range. Other factors also affect traits. A lack of sunshine or vital nutrients could stunt a plant's growth. How does genetics account for these issues? Mendel studied traits that follow simple dominant-recessive patterns of inheritance, and each trait was the result of a single gene. In general, however, inheritance is much more complex. Most alleles are not simply dominant or recessive; some are codominant. Many traits are influenced by multiple genes. The environment also interacts with genes and affects their expression. These complexities are discussed in Chapter 7.

**Contrast** Explain the difference between genotype and phenotype.

## 6.4 ASSESSMENT



### REVIEWING MAIN IDEAS

1. How are the terms **gene**, locus, and **allele** related?
2. Explain why an organism's genotype may be **homozygous** dominant, homozygous recessive, or **heterozygous**, but never heterozygous recessive.

### CRITICAL THINKING

3. **Apply** Suppose you are studying a fruit fly's DNA and you discover a gene for antenna length on chromosome 2. What word describes its location, and where would it be found in other fruit flies' DNA?
4. **Predict** If a **recessive** allele helps an organism reproduce, but the **dominant** allele hinders reproduction, which will be more common in a population?

### Connecting CONCEPTS

5. **Human Biology** Cystic fibrosis is a recessive disease that causes the production of abnormally thick, life-threatening mucus secretions. What is the **genotype** of a person with cystic fibrosis:  $CC$ ,  $Cc$ , or  $cc$ ? Explain.

## ▶ MAIN IDEA

# Genes influence the development of traits.

You may have heard about the Human Genome Project. Its goal was to find out the sequence of the 3 billion nucleotide pairs that make up a human's genome. A **genome** is all of an organism's genetic material. Unless you have an identical twin, you have a unique genome that determines all of your traits. Some of your traits can be seen, such as the color of your eyes. Other traits cannot be seen, such as the exact chemical makeup of your eyeball.

In genetics, we often focus on a single trait or set of traits. A genome is all of an organism's genes, but a **genotype** (JEHN-uh-TYP) typically refers to the genetic makeup of a specific set of genes. The genotype of a pea plant includes both of the genes that code for flower color, even if one of these genes is masked. In contrast, the physical characteristics, or traits, of an individual organism make up its **phenotype** (FEE-nuh-TYP). A pea plant with purple flowers has a phenotype for purple flowers. The plant might have a hidden gene for white flowers, but that does not matter to its phenotype.

## Dominant and Recessive Alleles

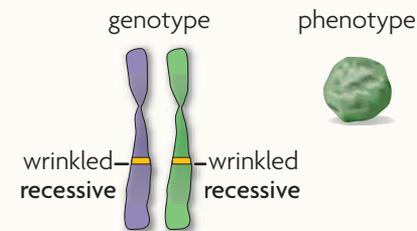
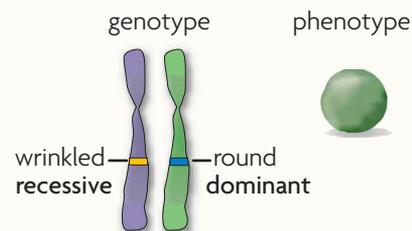
If an organism is heterozygous for a trait, which allele will be expressed? That is, if a plant has one allele for purple flowers and one for white flowers, what color will the flowers be? As Mendel learned, one allele may be dominant over another allele. A **dominant** allele is the allele that is expressed when two different alleles are present. A **recessive** allele is the allele that is only expressed when two copies are present. In Mendel's experiments, the allele for purple flowers was dominant to the allele for white flowers. All  $F_1$  plants were purple even though they had only one allele for purple flowers.

Sometimes the word *dominant* is misunderstood. A dominant allele is not necessarily better or stronger than a recessive allele. It does not necessarily occur most often in the population. An allele is dominant in a heterozygote simply because it is expressed and the other allele is not.

Alleles are often represented on paper with individual letters. An organism's genotype for a trait can be shown with two letters—one per allele. Uppercase letters are used for dominant alleles, and lowercase letters are used for recessive alleles. For example, the dominant allele for height in pea plants is written as *T*, for tall. The recessive allele for short plants is written as *t*.

## VISUAL VOCAB

A **dominant** allele is expressed when two different alleles are present.



A **recessive** allele is expressed only when two copies are present.



**FIGURE 6.11** Polydactyly is the condition of having more than the typical number of fingers or toes. The allele for polydactyly is dominant.

## Connecting CONCEPTS

**Exceptions to Mendel's Laws**  
Mendel's theory of inheritance cannot explain all patterns of inheritance. As you will learn in **Chapter 7**, incomplete dominance, codominance, polygenic traits, and environmental influences all provide exceptions.

# 6.5

## Traits and Probability

**KEY CONCEPT** The inheritance of traits follows the rules of probability.

### ▶ MAIN IDEAS

- Punnett squares illustrate genetic crosses.
- A monohybrid cross involves one trait.
- A dihybrid cross involves two traits.
- Heredity patterns can be calculated with probability.

### VOCABULARY

**Punnett square**, p. 183

**monohybrid cross**, p. 184

**testcross**, p. 185

**dihybrid cross**, p. 186

**law of independent assortment**, p. 186

**probability**, p. 187



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**Connect** If you have tried juggling, you know it can be a tricky thing. Keeping three flaming torches or juggling clubs in motion at the same time is a lot to handle. Trying to keep track of what organism has which genotype and which gamete gets which allele can also be a lot to juggle. Fortunately, R. C. Punnett developed a method to graphically keep track of all of the various combinations.

### ▶ MAIN IDEA

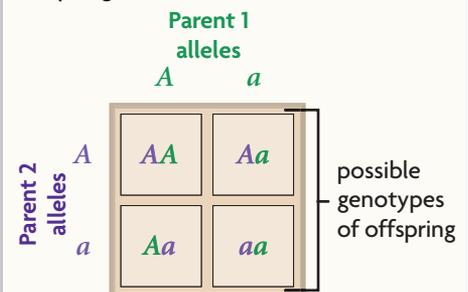
## Punnett squares illustrate genetic crosses.

Shortly after Mendel's experiments became widely known among scientists, a poultry geneticist named R. C. Punnett, shown in **FIGURE 6.13**, developed the Punnett square. A **Punnett square** is a grid system for predicting all possible genotypes resulting from a cross. The axes of the grid represent the possible gamete genotypes of each parent. The grid boxes show all of the possible genotypes of offspring from those two parents. Because segregation and fertilization are random events, each combination of alleles is as likely to be produced as any other. By counting the number of squares with each genetic combination, we can find the ratio of genotypes in that generation. If we also know how the genotype corresponds to the phenotype, we can find the ratio of phenotypes in that generation as well.

Let's briefly review what you've learned about meiosis and segregation to examine why the Punnett square is effective. Both parents have two alleles for each gene. These alleles are represented on the axes of the Punnett square. During meiosis, the chromosomes—and, therefore, the alleles—are separated.

### VISUAL VOCAB

The **Punnett square** is a grid system for predicting possible genotypes of offspring.



R. C. Punnett

**FIGURE 6.13** R. C. Punnett developed the Punnett square as a way to illustrate genetic crosses.

Each gamete gets one of the alleles. Since each parent contributes only one allele to the offspring, only one allele from each parent is written inside each grid box. Fertilization restores the diploid number in the resulting offspring, which is why each grid box has two alleles, one from the mother and one from the father. Since any egg has the same chance of being fertilized by any sperm cell, each possible genetic combination is equally likely to occur.

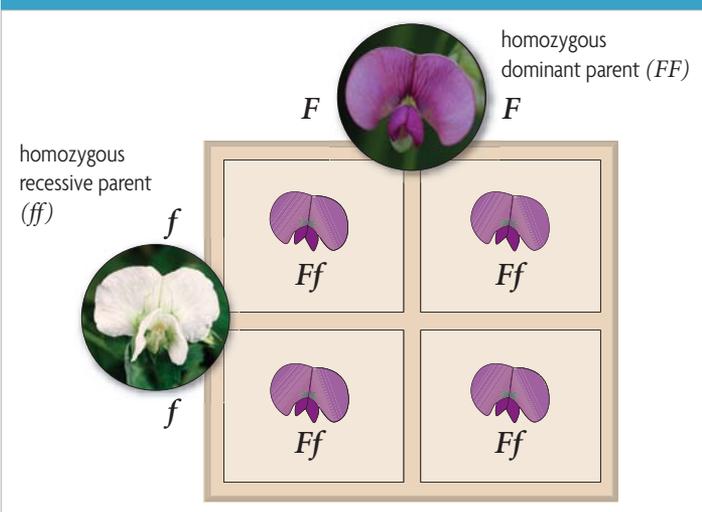
**Explain** What do the letters on the axes of the Punnett square represent?

**▶ MAIN IDEA**

## A monohybrid cross involves one trait.

Thus far, we have studied **monohybrid crosses**, crosses that examine the inheritance of only one specific trait. Three example crosses are used below and on the next page to illustrate how Punnett squares work and to highlight the resulting ratios—for both genotype and phenotype.

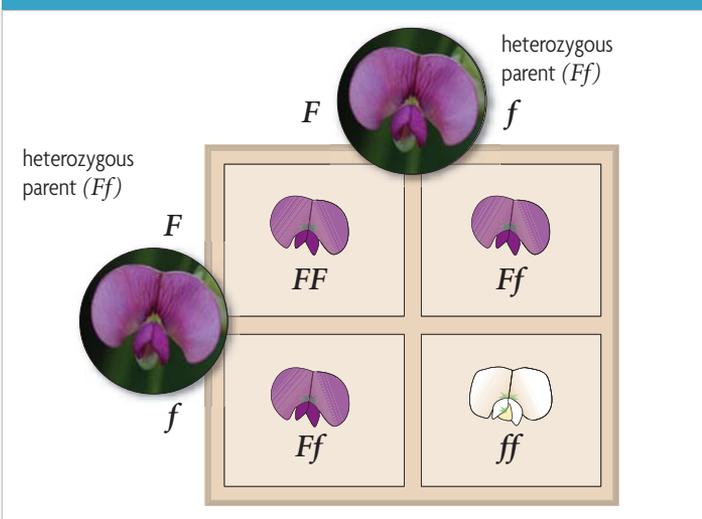
**FIGURE 6.14 HOMOZYGOUS-HOMOZYGOUS**



### Homozygous-Homozygous

Suppose you cross a pea plant that is homozygous dominant for purple flowers with a pea plant that is homozygous recessive for white flowers. To determine the genotypic and phenotypic ratios of the offspring, first write each parent's genotype on one axis:  $FF$  for the purple-flowered plant,  $ff$  for the white-flowered plant. Every gamete from the purple-flowered plant contains the dominant allele,  $F$ . Every gamete from the white-flowered plant contains the recessive allele,  $f$ . Therefore, 100 percent of the offspring have the heterozygous genotype,  $Ff$ . And 100 percent of the offspring have purple flowers, because they all have a copy of the dominant allele, as shown in **FIGURE 6.14**.

**FIGURE 6.15 HETEROZYGOUS-HETEROZYGOUS**



### Heterozygous-Heterozygous

Next, in **FIGURE 6.15**, you can see a cross between two purple-flowered pea plants that are both heterozygous ( $Ff$ ). From each parent, half the offspring receive a dominant allele,  $F$ , and half receive a recessive allele,  $f$ . Therefore, one-fourth of the offspring have a homozygous dominant genotype,  $FF$ ; half have a heterozygous genotype,  $Ff$ ; and one-fourth have a homozygous recessive genotype,  $ff$ . Both the  $FF$  and the  $Ff$  genotypes result in purple flowers. Only the  $ff$  genotype results in white flowers. Thus, the genotypic ratio is 1:2:1 of homozygous dominant:heterozygous:homozygous recessive. The phenotypic ratio is 3:1 of purple:white flowers.

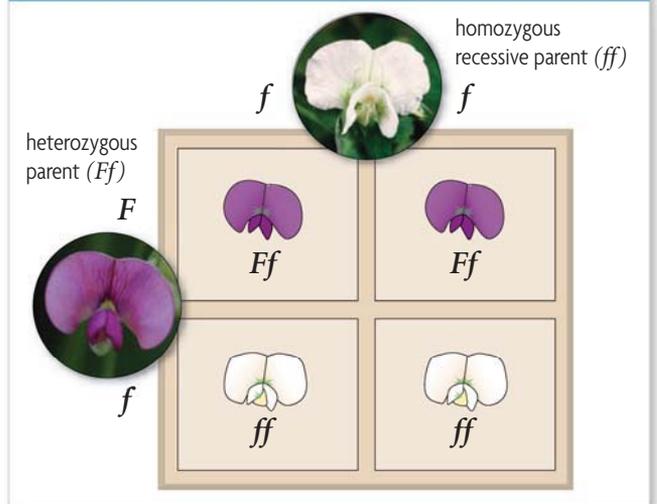
## Heterozygous-Homozygous

Finally, suppose you cross a pea plant that is heterozygous for purple flowers ( $Ff$ ) with a pea plant that is homozygous recessive for white flowers ( $ff$ ). As before, each parent's genotype is placed on an axis, as shown in **FIGURE 6.16**. From the homozygous parent with white flowers, the offspring each receive a recessive allele,  $f$ . From the heterozygous parent, half the offspring receive a dominant allele,  $F$ , and half receive a recessive allele,  $f$ . Half the offspring have a heterozygous genotype,  $Ff$ . Half have a homozygous recessive genotype,  $ff$ . Thus, half the offspring have purple flowers, and half have white flowers. The resulting genotypic ratio is 1:1 of heterozygous:homozygous recessive. The phenotypic ratio is 1:1 of purple:white.

Suppose we did not know the genotype of the purple flower in the cross above. This cross would allow us to determine that the purple flower is heterozygous, not homozygous dominant. A **testcross** is a cross between an organism with an unknown genotype and an organism with the recessive phenotype. The organism with the recessive phenotype must be homozygous recessive. The offspring will show whether the organism with the unknown genotype is heterozygous, as above, or homozygous dominant.

**Apply** From an  $FF \times Ff$  cross, what percent of offspring would have purple flowers?

**FIGURE 6.16 HETEROZYGOUS-HOMOZYGOUS**



## QUICK LAB INFERRING

### Using a Testcross

Suppose you work for a company that sells plant seeds. You are studying a plant species in which the dominant phenotype is pink flowers ( $PP$  or  $Pp$ ). The recessive phenotype is white flowers ( $pp$ ). Customers have been requesting more plants with pink flowers. To meet this demand, you need to determine the genotypes of some of the plants you are currently working with.

**PROBLEM** What is the genotype of each plant?

#### PROCEDURE

1. Suppose you are presented with Plant A of the species you are studying, which has pink flowers. You want to determine the genotype of the plant.
2. You cross Plant A with Plant B of the same species, which has white flowers and a known genotype of  $pp$ .
3. The resulting cross yields six plants with pink flowers and six plants with white flowers. Use Punnett squares to determine the genotype of Plant A.

#### ANALYZE AND CONCLUDE

1. **Apply** What is the genotype of Plant A? Explain how you arrived at your answer.
2. **Apply** What are the possible genotypes and phenotypes of offspring if Plant A is crossed with a plant that has a genotype of  $PP$ ?
3. **Calculate** What ratio of dominant to recessive phenotypes would exist if Plant A were crossed with a plant that has a genotype of  $pp$ ?
4. **Evaluate** Is Plant A the best plant, in terms of genotype, that you can work with to produce as many of the requested seeds as possible? Why or why not? Which genotype would be best to work with?

#### MATERIALS

- pencil
- paper

**MAIN IDEA**

## A dihybrid cross involves two traits.

### Connecting CONCEPTS

**Law of Segregation** As you learned in Section 6.3, Mendel's first law of inheritance is the law of segregation. It states that organisms inherit two copies of each gene but donate only one copy to each gamete.

All of the crosses discussed so far have involved only a single trait. However, Mendel also conducted **dihybrid crosses**, crosses that examine the inheritance of two different traits. He wondered if both traits would always appear together or if they would be expressed independently of each other.

Mendel performed many dihybrid crosses and tested a variety of different combinations. For example, he would cross a plant with yellow round peas with a plant with green wrinkled peas. Remember that Mendel began his crosses with purebred plants. Thus, the first generation offspring ( $F_1$ ) would all be heterozygous and would all look the same. In this example, the plants would all have yellow round peas. When Mendel allowed the  $F_1$  plants to self-pollinate, he obtained the following results: 9 yellow/round, 3 yellow/wrinkled, 3 green/round, 1 green/wrinkled.

Mendel continued to find this approximately 9:3:3:1 phenotypic ratio in the  $F_2$  generation, regardless of the combination of traits. From these results, he realized that the presence of one trait did not affect the presence of another trait. His second law of genetics, the **law of independent assortment**, states that allele pairs separate independently of each other during gamete formation, or meiosis. That is, different traits appear to be inherited separately.

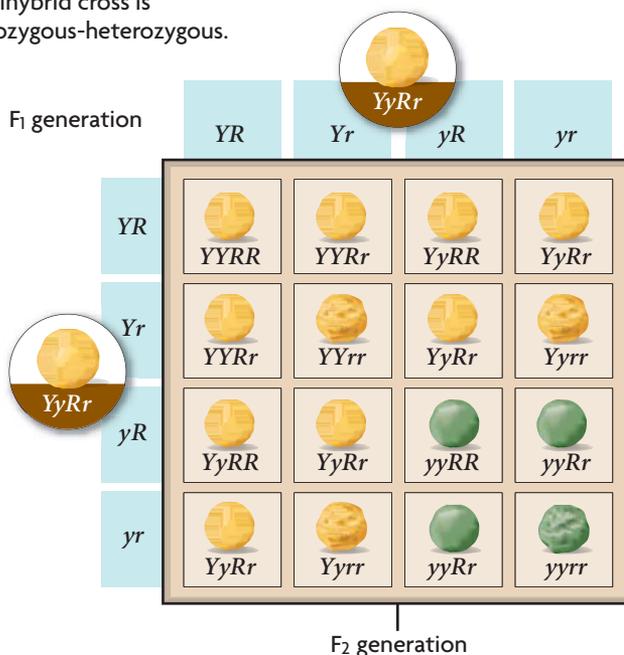
The results of Mendel's dihybrid crosses can also be illustrated with a Punnett square, like the one in **FIGURE 6.17**. Drawing a Punnett square for a dihybrid cross is the same as drawing one for a monohybrid cross, except that the grid is bigger because two genes, or four alleles, are involved. For example, suppose you cross two plants with yellow, round peas that are heterozygous for both traits ( $YyRr$ ). The four allele combinations possible in each gamete—

$YR$ ,  $Yr$ ,  $yR$ , and  $yr$ —are used to label each axis. Each grid box can be filled in using the same method as that used in the monohybrid cross. A total of nine different genotypes may result from the cross in this example. However, these nine genotypes produce only four different phenotypes. These phenotypes are yellow round, yellow wrinkled, green round, and green wrinkled, and they occur in the ratio of 9:3:3:1. Note that the 9:3:3:1 phenotypic ratio results from a cross between organisms that are heterozygous for both traits. The phenotypic ratio of the offspring will differ (from 9:3:3:1) if one or both of the parent organisms are homozygous for one or both traits.

**Analyze** In **FIGURE 6.17**, the boxes on the axes represent the possible gametes made by each parent plant. Why does each box have two alleles?

**FIGURE 6.17 DIHYBRID CROSS**

This dihybrid cross is heterozygous-heterozygous.



## MAIN IDEA

# Heredity patterns can be calculated with probability.

**Probability** is the likelihood that a particular event will happen. It predicts the average number of occurrences, not the exact number of occurrences.

$$\text{Probability} = \frac{\text{number of ways a specific event can occur}}{\text{number of total possible outcomes}}$$

Suppose you flip a coin. The number of total possible outcomes is two: heads up or tails up. The probability that it would land heads up is  $1/2$ , or one out of two. The probability that it would land tails up is also  $1/2$ .

Next, suppose you flip two coins. How one coin lands does not affect how the other coin lands. To calculate the probability that two independent events will happen together, multiply the probability of each individual event. The probability that both coins will land heads up, for example, is  $1/2 \times 1/2 = 1/4$ .

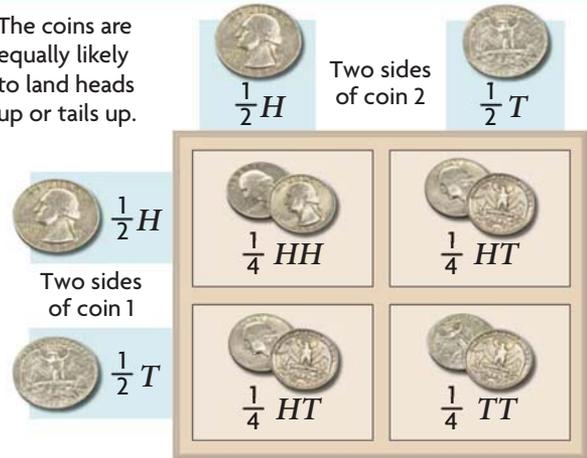
These probabilities can be applied to meiosis. Suppose a germ cell undergoes meiosis in a plant that is heterozygous for purple flowers. The number of total possible outcomes is two because a gamete could get a dominant or a recessive allele. The probability that a gamete will get a dominant allele is  $1/2$ . The probability that it will get a recessive allele is also  $1/2$ .

If two plants that are heterozygous for purple flowers fertilize each other, the probability that both egg and sperm have a dominant allele is  $1/2 \times 1/2 = 1/4$ . So, too, the probability that both have a recessive allele is  $1/4$ . There is also a  $1/4$  chance that a sperm cell with a dominant allele will fertilize an egg with a recessive allele, or that a sperm cell with a recessive allele will fertilize an egg with a dominant allele. These last two combinations are basically the same. In either case, the resulting plant will be heterozygous. Thus, the probability that a pea plant will be heterozygous for this trait is the sum of the probabilities:  $1/4 + 1/4 = 1/2$ .

**Apply** Explain how Mendel's laws relate to probability.

FIGURE 6.18 PROBABILITY AND HEREDITY

The coins are equally likely to land heads up or tails up.



## 6.5 ASSESSMENT

ONLINE QUIZ  
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### REVIEWING MAIN IDEAS

1. What do the grid boxes in a **Punnett square** represent?
2. Why does the expected genotypic ratio often differ from the expected phenotypic ratio resulting from a **monohybrid cross**?
3. How did Mendel's **dihybrid crosses** help him develop his second law?

### CRITICAL THINKING

4. **Calculate** What would be the phenotypic ratios of the offspring resulting from the following cross:  $YYRr \times YyRr$ ?
5. **Predict** If you are working with two tall pea plants and know that one is  $Tt$ , how could you determine the genotype of the other plant?

### Connecting CONCEPTS

6. **Adaptation** You have seen that one-quarter of offspring resulting from two heterozygous parents are homozygous recessive. Yet for some genes, the recessive allele is more common in the population. Explain why this might be.