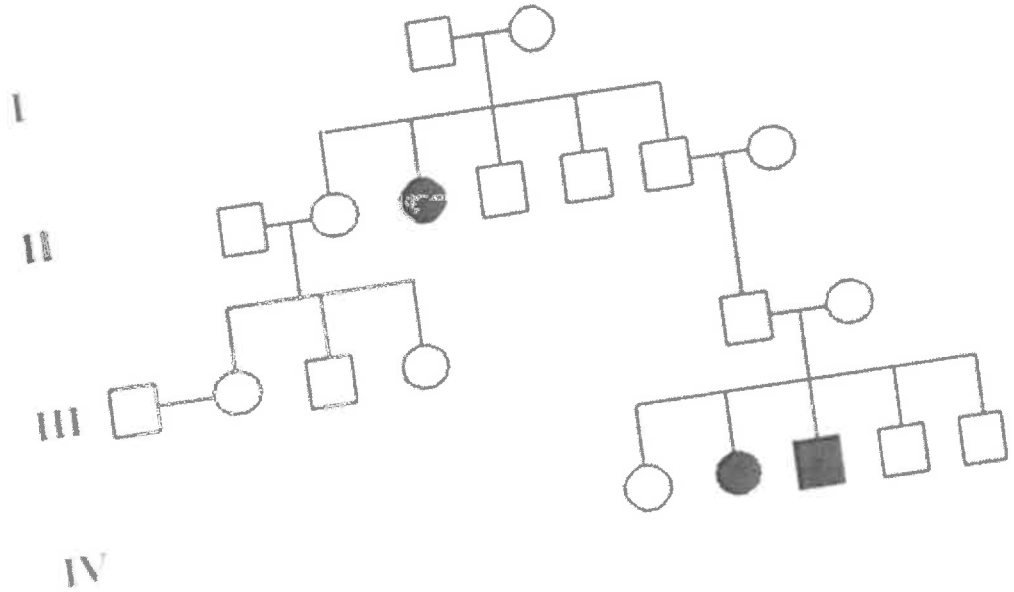
GRADE 12 BIOLOGY

REFERENCE READING PACKAGE

UNIT A – GENETICS



Revised 2016-08-30

10.1 Cell Growth, Division, and Reproduction

Key Questions

What are some of the difficulties a cell faces as it increases in size?

How do asexual and sexual reproduction compare?

Vocabulary

cell division
asexual reproduction
sexual reproduction

Taking Notes

Outline As you read, create an outline about cell growth, division, and reproduction. As you read, fill in key phrases or sentences about each heading.

THINK ABOUT IT When a living thing grows, what happens to its cells? Does an organism get larger because each cell increases in size or because it produces more of them? In most cases, living things grow by producing more cells. What is there about growth that requires cells to divide and produce more of themselves?

Limits to Cell Size

What are some of the difficulties a cell faces as it increases in size?

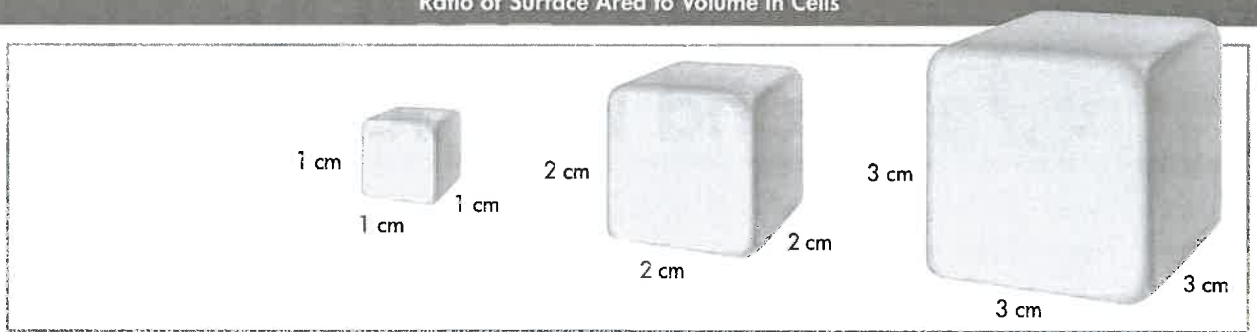
Nearly all cells can grow by increasing in size, but eventually, most cells divide after growing to a certain point. There are two main reasons why cells divide rather than continuing to grow. **The larger a cell becomes, the more demands the cell places on its DNA. In addition, a larger cell is less efficient in moving nutrients and waste materials across the cell membrane.**

Information "Overload" Living cells store critical information in a molecule known as DNA. As a cell grows, that information is used to build the molecules needed for cell growth. But as a cell increases in size, its DNA does not. If a cell were to grow too large, an "information crisis" would occur.

To get a better sense of information overload, compare a cell to a growing town. Suppose a small town has a library with a few thousand books. As more people move in, more people will borrow books. Sometimes, people may have to wait to borrow popular books. Similarly, a larger cell would make greater demands on its genetic "library." After a while, the DNA would no longer be able to serve the needs of the growing cell—it might be time to build a new library.

Exchanging Materials There is another critical reason why cell size is limited. Food, oxygen, and water enter a cell through its cell membrane. Waste products leave a cell in the same way. The rate at which this exchange takes place depends on the surface area of the cell, which is the total area of its cell membrane. The rate at which food and oxygen are used up and waste products are produced depends on the cell's volume. Understanding the relationship between a cell's surface area and its volume is the key to understanding why cells must divide rather than continue to grow.

Ratio of Surface Area to Volume in Cells

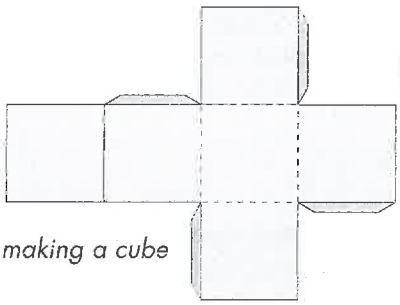


Surface Area (length x width) x 6 sides	1 cm x 1 cm x 6 = 6 cm^2	2 cm x 2 cm x 6 = 24 cm^2	3 cm x 3 cm x 6 = 54 cm^2
Volume (length x width x height)	1 cm x 1 cm x 1 cm = 1 cm^3	2 cm x 2 cm x 2 cm = 8 cm^3	3 cm x 3 cm x 3 cm = 27 cm^3
Ratio of Surface Area to Volume	6 / 1 = 6 : 1	24 / 8 = 3 : 1	54 / 27 = 2 : 1

FIGURE 10–1 Ratio of Surface Area to Volume As the length of the sides increases, the volume increases more than the surface area. **Interpret Tables** ***What are the ratios comparing?***

▶ ***Ratio of Surface Area to Volume*** Imagine a cell that is shaped like a cube, like those shown in **Figure 10–1**. The formula for area ($l \times w$) is used to calculate the surface area. The formula for volume ($l \times w \times h$) is used to calculate the amount of space inside. By using a ratio of surface area to volume, you can see how the size of the cell's surface area grows compared to its volume.

Notice that for a cell with sides that measure 1 cm in length, the ratio of surface area to volume is 6/1 or 6 : 1. Increase the length of the cell's sides to 2 cm, and the ratio becomes 24/8 or 3 : 1. What if the length triples? The ratio of surface area to volume becomes 54/27 or 2 : 1. Notice that the surface area is not increasing as fast as the volume increases. For a growing cell, a decrease in the relative amount of cell membrane available creates serious problems.



making a cube

Quick Lab
OPEN-ENDED INQUIRY

Modeling the Relationship Between Surface Area and Volume

1. Use the drawing and grid paper to make patterns for a 6-cm cube, a 5-cm cube, a 4-cm cube, and a 3-cm cube.
2. Cut out your patterns and fold them. Then use the tabs to tape or glue the sides together. Don't tape down the top side.
3. Construct a data table to compare the volume, the surface area, and the ratio of surface area to volume of each cube.
4. Use your data to calculate the number of 3-cm cubes that would fit in the same volume as the 6-cm cube. Also calculate the total surface area for the smaller cubes.

Analyze and Conclude

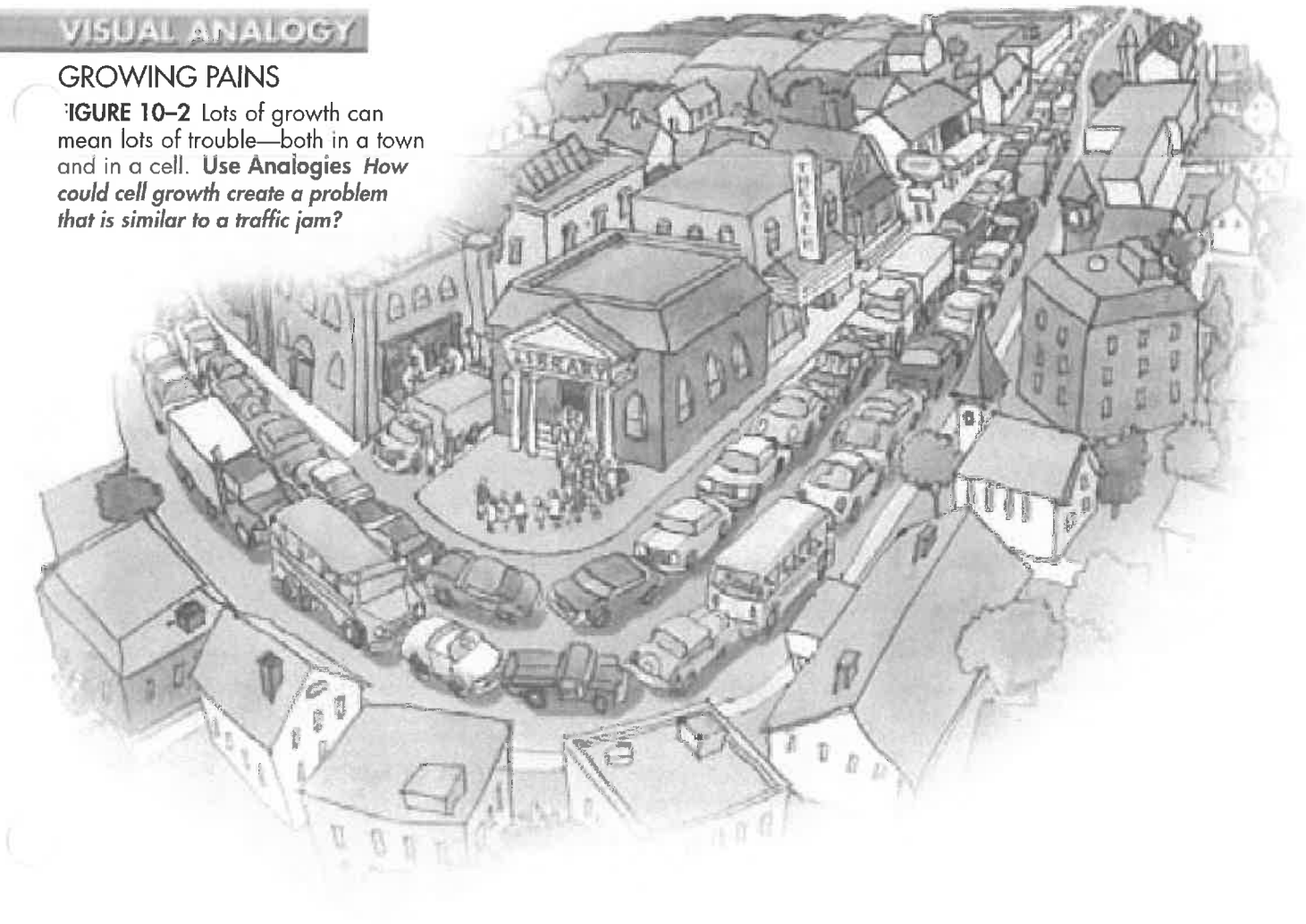
1. Review Describe the function of a cell membrane and its relationship to what happens inside a cell.

2. Apply Concepts How does the surface area change when a large cell divides into smaller cells that have the same total volume?

VISUAL ANALOGY

GROWING PAINS

FIGURE 10–2 Lots of growth can mean lots of trouble—both in a town and in a cell. **Use Analogies** ***How could cell growth create a problem that is similar to a traffic jam?***



▶ ***Traffic Problems*** To use the town analogy again, suppose the town has just a two-lane main street leading to the center of town. As the town grows, more and more traffic clogs the main street. It becomes increasingly difficult to move goods in and out.

A cell that continues to grow would experience similar problems. If a cell got too large, it would be more difficult to get sufficient amounts of oxygen and nutrients in and waste products out. This is another reason why cells do not continue to grow larger even if the organism does.

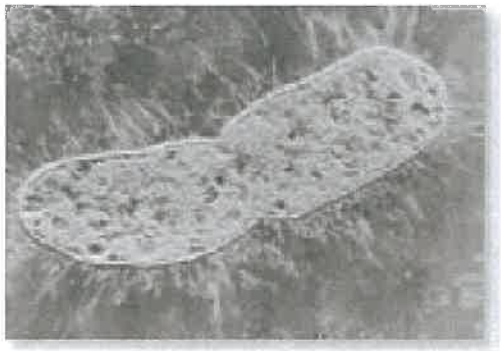
Division of the Cell Before it becomes too large, a growing cell divides, forming two "daughter" cells. The process by which a cell divides into two new daughter cells is called **cell division.**

Before cell division occurs, the cell replicates, or copies all of its DNA. This replication of DNA solves the problem of information overload because each daughter cell gets one complete copy of genetic information. Cell division also solves the problem of increasing size by reducing cell volume. Cell division results in an increase in the ratio of surface area to volume for each daughter cell. This allows for the efficient exchange of materials within a cell.

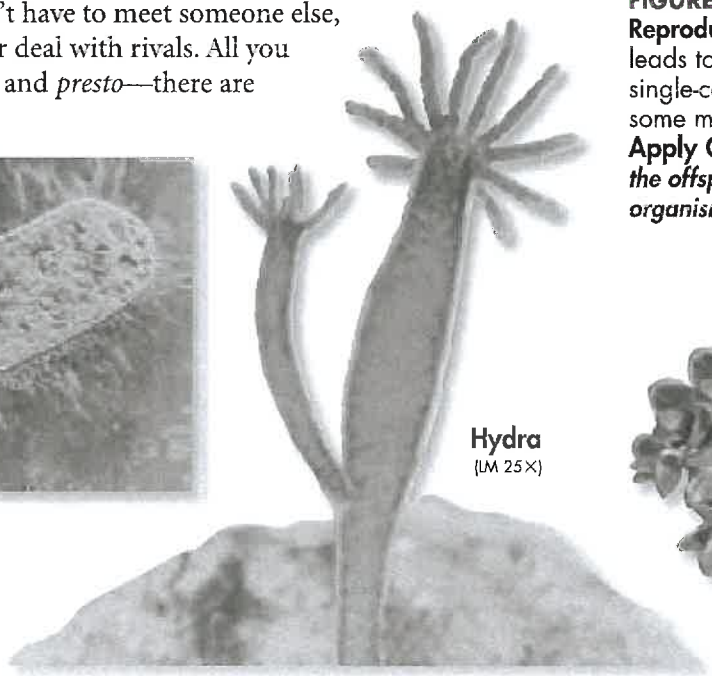
Cell Division and Reproduction

How do asexual and sexual reproduction compare?

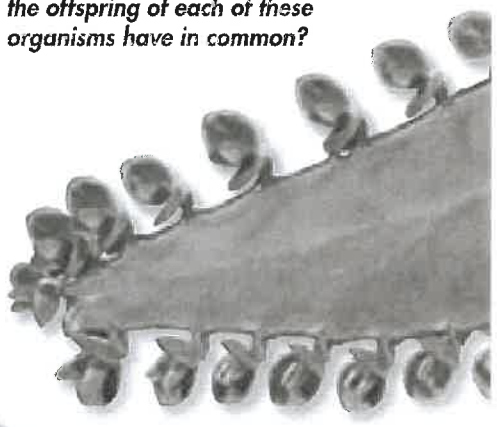
Reproduction, the formation of new individuals, is one of the most important characteristics of living things. For an organism composed of just one cell, cell division can serve as a perfectly good form of reproduction. You don't have to meet someone else, conduct a courtship, or deal with rivals. All you have to do is to divide, and *presto*—there are two of you!



Bacterium
(TEM 32,800×)



Hydra
(LM 25×)



Kalanchoe

FIGURE 10–3 Asexual Reproduction Cell division leads to reproduction in single-celled organisms and some multicellular organisms. **Apply Concepts** ***What do the offspring of each of these organisms have in common?***

Asexual Reproduction For many single-celled organisms, such as the bacterium in **Figure 10–3**, cell division is the only form of reproduction. The process can be relatively simple, efficient, and effective, enabling populations to increase in number very quickly. In most cases, the two cells produced by cell division are genetically identical to the cell that produced them. This kind of reproduction is called **asexual reproduction.** **The production of genetically identical offspring from a single parent is known as asexual reproduction.**

Asexual reproduction also occurs in many multicellular organisms. The small bud growing off the hydra will eventually break off and become an independent organism, an example of asexual reproduction in an animal. Each of the small shoots or plantlets on the tip of the kalanchoe leaf may also grow into a new plant.

Sexual Reproduction Unlike asexual reproduction, where cells separate to form a new individual, **sexual reproduction** involves the fusion of two separate parent cells. In sexual reproduction, offspring are produced by the fusion of special reproductive cells formed by each of two parents. **Offspring produced by sexual reproduction inherit some of their genetic information from each parent.** Most animals and plants reproduce sexually, and so do some single-celled organisms. You will learn more about the form of cell division that produces reproductive cells in Chapter 11.

In Your Notebook *Use a Venn diagram to compare asexual and sexual reproduction.*

BUILD Vocabulary

PREFIXES The prefix *a-* in *asexual* means "without." **Asexual reproduction** is reproduction without the fusion of reproductive cells.

MYSTERY CLUE

As its wound heals, the salamander's body cells are dividing to repair the damage. In what way is this type of cell division similar to asexual reproduction?

Comparing Asexual and Sexual Reproduction You can see that each type of reproduction has its advantages and disadvantages when you look at each one as a strategy for survival. Species survive by reproducing. The better suited a species is to its environment, the greater its chance of survival.

For single-celled organisms, asexual reproduction is a survival strategy. When conditions are right, the faster they reproduce, the better their chance of survival over other organisms using the same resources. Having offspring that are genetically identical is also an advantage as long as conditions remain favorable. However, a lack of genetic diversity becomes a disadvantage when conditions change in ways that do not fit the characteristics of an organism.

Sexual reproduction is a different type of survival strategy. The process of finding a mate and the growth and development of offspring require more time. However, this can be an advantage for species that live in environments where seasonal changes affect weather conditions and food availability. Sexual reproduction also provides genetic diversity. If an environment changes, some offspring may have the right combination of characteristics needed to survive.

Some organisms reproduce both sexually and asexually. Yeasts, for example, are single-celled eukaryotes that use both strategies. They reproduce asexually most of the time. However, under certain conditions, they enter a sexual phase. The different advantages of each type of reproduction may help to explain why the living world includes organisms that reproduce sexually, those that reproduce asexually, and many organisms that do both.

10.1 Assessment

Review Key Concepts

1. a. Review Identify two reasons why a cell's growth is limited.

b. Explain As a cell's size increases, what happens to the ratio of its surface area to its volume?

c. Applying Concepts Why is a cell's surface area-to-volume ratio important?

2. a. Review What is asexual reproduction? What is sexual reproduction?

b. Explain What types of organisms reproduce sexually?

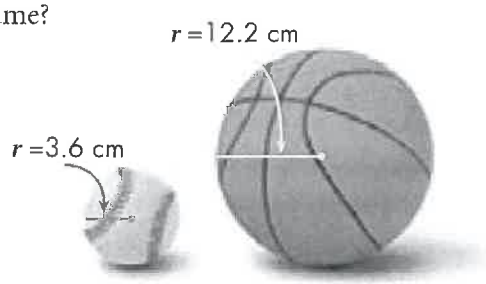
c. Summarize What are the advantages and disadvantages of both asexual and sexual reproduction?

VISUAL THINKING MATH

3. The formula for finding the surface area of a sphere, such as a baseball or a basketball, is $A = 4\pi r^2$, where r is the radius. The formula for finding the volume of a sphere is $V = 4/3\pi r^3$.

a. Calculate Calculate the surface area and the volume of the baseball and the basketball. Then, write the ratio of surface area to volume for each sphere.

b. Infer If the baseball and basketball were cells, which would possess a larger ratio of area of cell membrane to cell volume?



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10.2 The Process of Cell Division

THINK ABOUT IT What role does cell division play in your life? You know from your own experience that living things grow, or increase in size, during particular stages of life or even throughout their lifetime. This growth clearly depends on the production of new cells through cell division. But what happens when you are finished growing? Does cell division simply stop? Think about what must happen when your body heals a cut or a broken bone. And finally, think about the everyday wear and tear on the cells of your skin, digestive system, and blood. Cell division has a role to play there, too.

Chromosomes

What is the role of chromosomes in cell division?

What do you think would happen if a cell were simply to split in two, without any advance preparation? The results might be disastrous, especially if some of the cell's essential genetic information wound up in one of the daughter cells, and not in the other. In order to make sure this doesn't happen, cells first make a complete copy of their genetic information before cell division begins.

Even a small cell like the bacterium *E. coli* has a tremendous amount of genetic information in the form of DNA. In fact, the total length of this bacterium's DNA molecule is 1.6 mm, roughly 1000 times longer than the cell itself. In terms of scale, imagine a 300-meter rope stuffed into a school backpack. Cells can handle such large molecules only by careful packaging. Genetic information is bundled into packages of DNA known as **chromosomes.**

Prokaryotic Chromosomes Prokaryotes lack nuclei and many of the organelles found in eukaryotes. Their DNA molecules are found in the cytoplasm along with most of the other contents of the cell. Most prokaryotes contain a single, circular DNA chromosome that contains all, or nearly all, of the cell's genetic information.

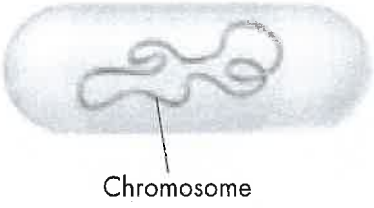


FIGURE 10–4 Prokaryotic Chromosome In most prokaryotes, a single chromosome holds most of the organism's DNA.

Key Questions

- ***What is the role of chromosomes in cell division?***
- ***What are the main events of the cell cycle?***
- ***What events occur during each of the four phases of mitosis?***
- ***How do daughter cells split apart after mitosis?***

Vocabulary

chromosome • chromatin • cell cycle • interphase • mitosis • cytokinesis • prophase • centromere • chromatid • centriole • metaphase • anaphase • telophase

Taking Notes

Two-Column Chart As you read, create a two-column chart. In the left column, make notes about what is happening in each stage of the cell cycle. In the right column, describe what the process looks like or draw pictures.

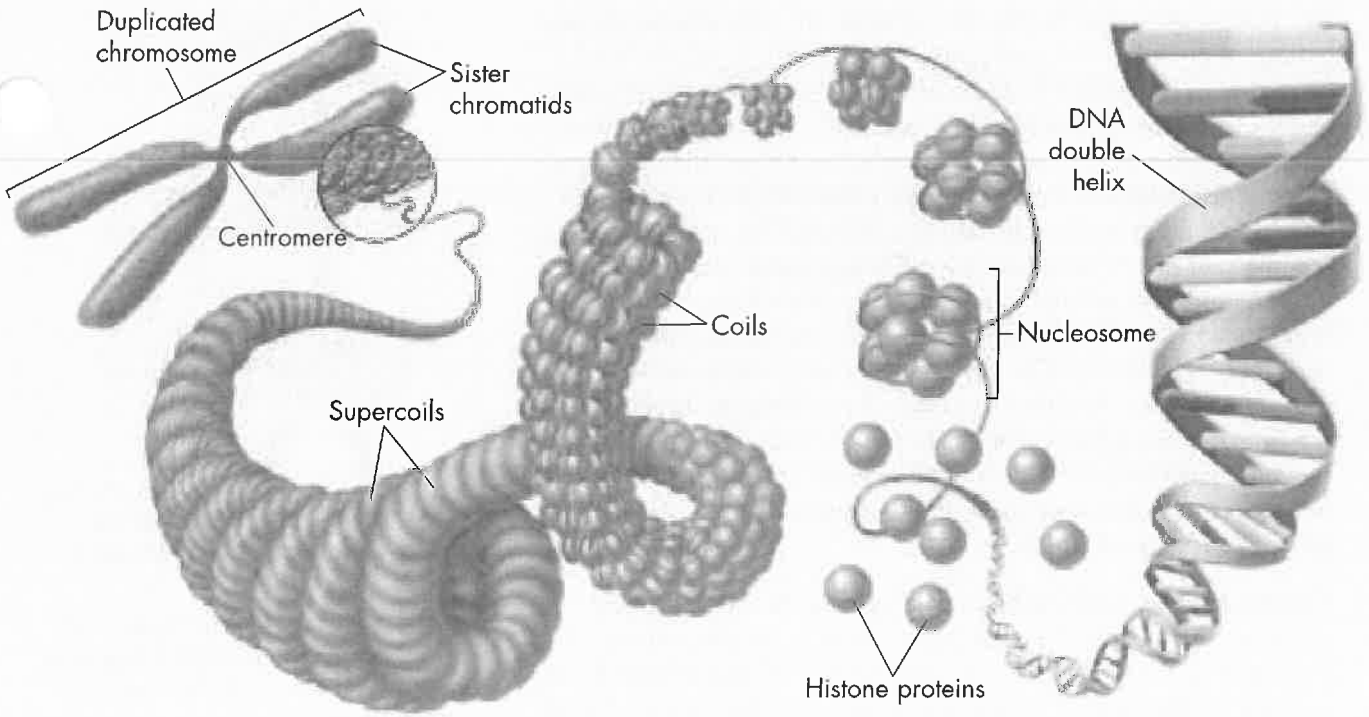


FIGURE 10–5 Eukaryotic Chromosome As a eukaryotic cell prepares for division, each chromosome coils more and more tightly to form a compact structure. **nterpret Visuals** *Which side of the diagram, left or right, shows the smallest structures, and which shows the largest?*

Eukaryotic Chromosomes Eukaryotic cells generally have much more DNA than prokaryotes have and, therefore, contain multiple chromosomes. Fruit flies, for example, have 8 chromosomes per cell, human cells have 46, and carrot cells have 18. The chromosomes in eukaryotic cells form a close association with histones, a type of protein. This complex of chromosome and protein is referred to as **chromatin.** DNA tightly coils around the histones, and together, the DNA and histone molecules form beadlike structures called nucleosomes. Nucleosomes pack together to form thick fibers, which condense even further during cell division. Usually the chromosome shape you see drawn is a duplicated chromosome with supercoiled chromatin, as shown in **Figure 10–5.**

Why do cells go to such lengths to package their DNA into chromosomes? One of the principal reasons is to ensure equal division of DNA when a cell divides. **Chromosomes make it possible to separate DNA precisely during cell division.**

In Your Notebook *Write instructions to build a eukaryotic chromosome.*

The Cell Cycle

What are the main events of the cell cycle?

Cells go through a series of events known as the **cell cycle** as they grow and divide. **During the cell cycle, a cell grows, prepares for division, and divides to form two daughter cells.** Each daughter cell then moves into a new cell cycle of activity, growth, and division.

The Prokaryotic Cell Cycle The prokaryotic cell cycle is a regular pattern of growth, DNA replication, and cell division that can take place very rapidly under ideal conditions. Researchers are only just beginning to understand how the cycle works in prokaryotes, and relatively little is known about its details. It is known that most prokaryotic cells begin to replicate, or copy, their DNA chromosomes once they have grown to a certain size. When DNA replication is complete, or nearly complete, the cell begins to divide.

The process of cell division in prokaryotes is a form of asexual reproduction known as binary fission. Once the chromosome has been replicated, the two DNA molecules attach to different regions of the cell membrane. A network of fibers forms between them, stretching from one side of the cell to the other. The fibers constrict and the cell is pinched inward, dividing the cytoplasm and chromosomes between two newly formed cells. Binary fission results in the production of two genetically identical cells.

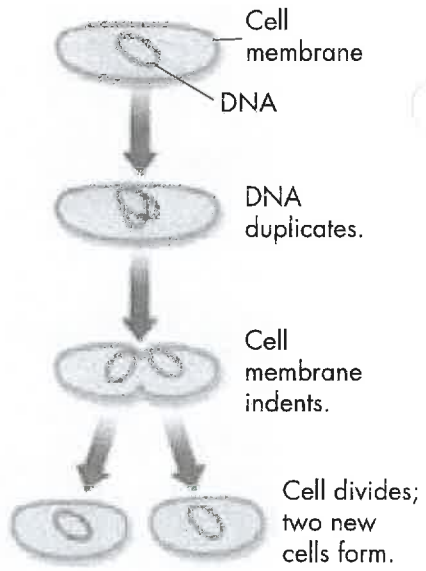


FIGURE 10–6 Binary Fission Cell division in a single-celled organism produces two genetically identical organisms.

The Eukaryotic Cell Cycle In contrast to prokaryotes, much more is known about the eukaryotic cell cycle. As you can see in **Figure 10–7,** the eukaryotic cell cycle consists of four phases: G_1, S, G_2, and M. The length of each part of the cell cycle—and the length of the entire cell cycle—varies depending on the type of cell.

At one time, biologists described the life of a cell as one cell division after another separated by an "in-between" period of growth called **interphase.** We now appreciate that a great deal happens in the time between cell divisions. Interphase is divided into three parts: G_1, S, and G_2.

▶ ***G_1 Phase: Cell Growth*** Cells do most of their growing during the G_1 phase. In this phase, cells increase in size and synthesize new proteins and organelles. The *G* in G_1 and G_2 stands for "gap," but the G_1 and G_2 phases are actually periods of intense growth and activity.

▶ ***S Phase: DNA Replication*** The G_1 phase is followed by the S phase. The *S* stands for "synthesis." During the S phase, new DNA is synthesized when the chromosomes are replicated. The cell at the end of the S phase contains twice as much DNA as it did at the beginning.

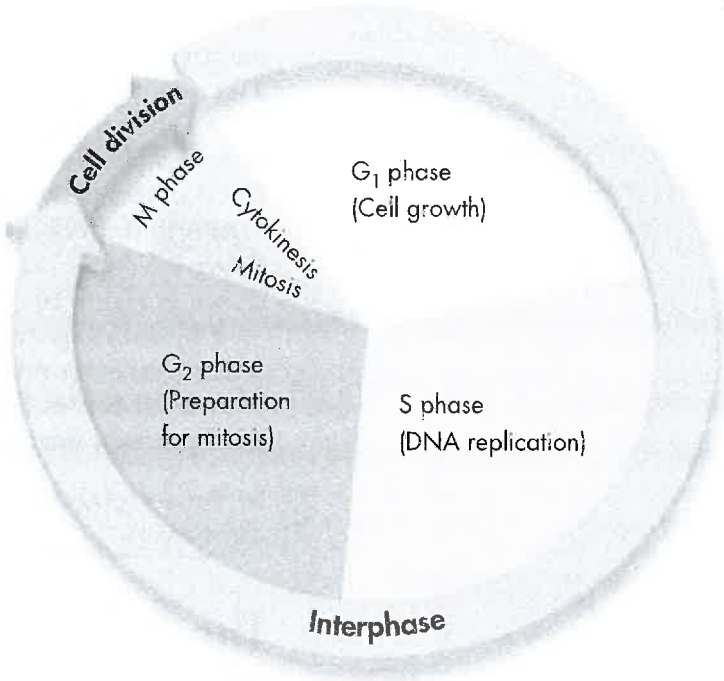


FIGURE 10–7 The Cell Cycle During the cell cycle, a cell grows, prepares for division, and divides to form two daughter cells. The cell cycle includes four phases—G_1, S, G_2, and M. **Infer** ***During which phase or phases would you expect the amount of DNA in the cell to change?***

▶ ***G_2 Phase: Preparing for Cell Division*** When DNA replication is completed, the cell enters the G_2 phase. G_2 is usually the shortest of the three phases of interphase. During the G_2 phase, many of the organelles and molecules required for cell division are produced. When the events of the G_2 phase are completed, the cell is ready to enter the M phase and begin the process of cell division.

▶ ***M Phase: Cell Division*** The M phase of the cell cycle, which follows interphase, produces two daughter cells. The M phase takes its name from the process of mitosis. During the normal cell cycle, interphase can be quite long. In contrast, the process of cell division usually takes place quickly.

In eukaryotes, cell division occurs in two main stages. The first stage of the process, division of the cell nucleus, is called **mitosis** (my TOH sis). The second stage, the division of the cytoplasm, is called **cytokinesis** (sy toh kih NEE sis). In many cells, the two stages may overlap, so that cytokinesis begins while mitosis is still taking place.

BUILD Vocabulary

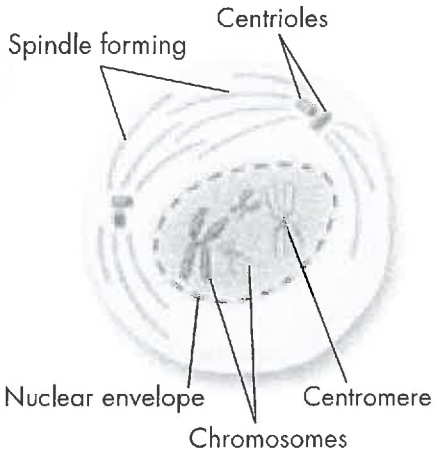
WORD ORIGINS The prefix *cyto-* in **cytokinesis** refers to cells and derives from the Greek word *kytos*, meaning "a hollow vessel." *Cytoplasm* is another word that has the same root.

Mitosis

What events occur during each of the four phases of mitosis?

Biologists divide the events of mitosis into four phases: prophase, metaphase, anaphase, and telophase. Depending on the type of cell, mitosis may last anywhere from a few minutes to several days. **Figure 10–8** through **Figure 10–11** show mitosis in an animal cell.

FIGURE 10–8 Prophase

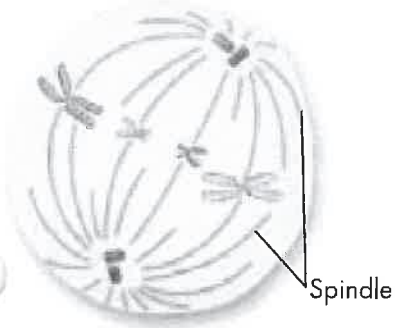


Prophase The first phase of mitosis, **prophase,** is usually the longest and may take up to half of the total time required to complete mitosis. **During prophase, the genetic material inside the nucleus condenses and the duplicated chromosomes become visible. Outside the nucleus, a spindle starts to form.**

The duplicated strands of the DNA molecule can be seen to be attached along their length at an area called the **centromere.** Each DNA strand in the duplicated chromosome is referred to as a **chromatid** (KROH muh tid), or sister chromatid. When the process of mitosis is complete, the chromatids will have separated and been divided between the new daughter cells.

Also during prophase, the cell starts to build a spindle, a fanlike system of microtubules that will help to separate the duplicated chromosomes. Spindle fibers extend from a region called the centrosome, where tiny paired structures called **centrioles** are located. Plant cells lack centrioles, and organize spindles directly from their centrosome regions. The centrioles, which were duplicated during interphase, start to move toward opposite ends, or poles, of the cell. As prophase ends, the chromosomes coil more tightly, the nucleolus disappears, and the nuclear envelope breaks down.

FIGURE 10–9 Metaphase



Metaphase The second phase of mitosis, **metaphase,** is generally the shortest. **During metaphase, the centromeres of the duplicated chromosomes line up across the center of the cell. Spindle fibers connect the centromere of each chromosome to the two poles of the spindle.**

Anaphase The third phase of mitosis, **anaphase,** begins when sister chromatids suddenly separate and begin to move apart. Once anaphase begins, each sister chromatid is now considered an individual chromosome. **During anaphase, the chromosomes separate and move along spindle fibers to opposite ends of the cell.** Anaphase comes to an end when this movement stops and the chromosomes are completely separated into two groups.

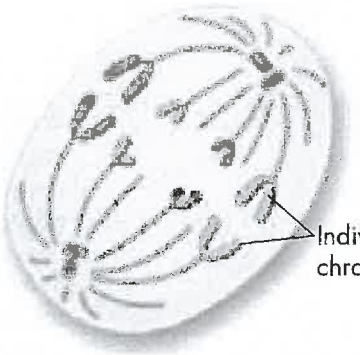


FIGURE 10–10
Anaphase

Telophase Following anaphase is **telophase,** the fourth and final phase of mitosis. **During telophase, the chromosomes, which were distinct and condensed, begin to spread out into a tangle of chromatin.** A nuclear envelope re-forms around each cluster of chromosomes. The spindle begins to break apart, and a nucleolus becomes visible in each daughter nucleus. Mitosis is complete. However, the process of cell division has one more step to go.

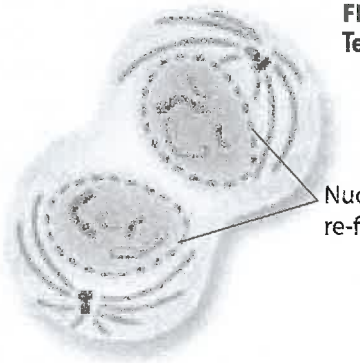


FIGURE 10–11
Telophase

In Your Notebook *Create a chart that lists the important information about each phase of mitosis.*

Quick Lab
GUIDED INQUIRY

Mitosis in Action

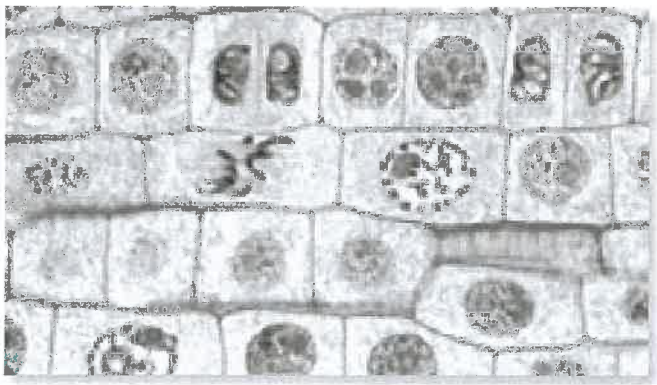
1. Examine a slide of a stained onion root tip under a microscope. Viewing the slide under low power, adjust the stage until you find the boxlike cells just above the root tip.
2. Switch the microscope to high power and locate cells that are in the process of dividing.
3. Find and sketch cells that are in each phase of mitosis. Label each sketch with the name of the appropriate phase.

Analyze and Conclude

1. Observe In which phase of the cell cycle were most of the cells you observed? Why do you think this is?

2. Draw Conclusions What evidence did you observe that shows mitosis is a continuous process, not a series of separate events?

3. Apply Concepts Cells in the root divide many times as the root grows longer and thicker. With each cell division, the chromosomes are divided between two daughter cells, yet the number of chromosomes in each cell does not change. What processes ensure that the normal number of chromosomes is restored after each cell division?



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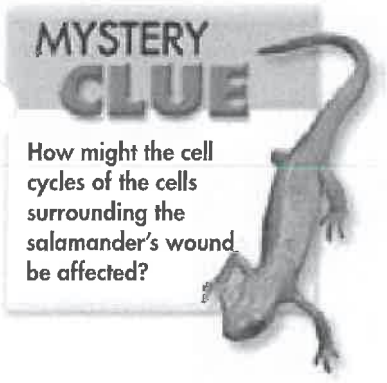


Cytokinesis

How do daughter cells split apart after mitosis?

As a result of mitosis, two nuclei—each with a duplicate set of chromosomes—are formed. All that remains to complete the M phase of the cycle is cytokinesis, the division of the cytoplasm itself. Cytokinesis usually occurs at the same time as telophase. **Cytokinesis completes the process of cell division—it splits one cell into two.** The process of cytokinesis differs in animal and plant cells.

Cytokinesis in Animal Cells During cytokinesis in most animal cells, the cell membrane is drawn inward until the cytoplasm is pinched into two nearly equal parts. Each part contains its own nucleus and cytoplasmic organelles.

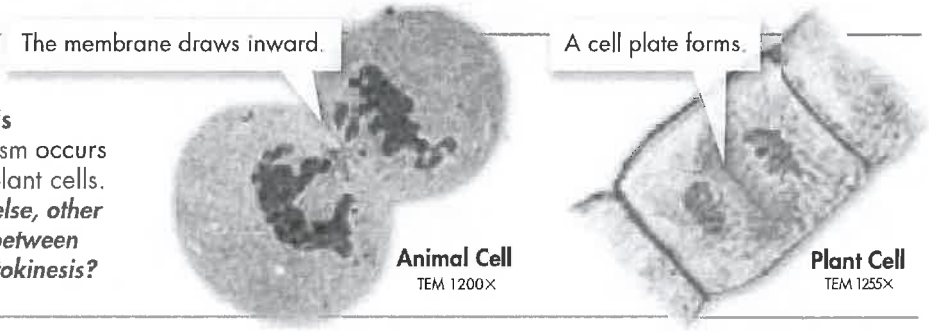


FIGURE 10–12 Cytokinesis
The division of the cytoplasm occurs differently in animal and plant cells.
Draw Conclusions ***What else, other than cytoplasm, is divided between the two new cells during cytokinesis?***

Cytokinesis in Plant Cells Cytokinesis in plant cells proceeds differently. The cell membrane is not flexible enough to draw inward because of the rigid cell wall that surrounds it. Instead, a structure known as the cell plate forms halfway between the divided nuclei. The cell plate gradually develops into cell membranes that separate the two daughter cells. A cell wall then forms in between the two new membranes, completing the process.

10.2 Assessment

Review Key Concepts

1. **a. Review** What are chromosomes?
   b. Compare and Contrast How does the structure of chromosomes differ in prokaryotes and eukaryotes?
2. **a. Review** What is the cell cycle?
   b. Sequence During which phase of the cell cycle are chromosomes replicated?
3. **a. Review** What happens during each of the four phases of mitosis? Write one or two sentences for each phase.
   b. Predict What do you predict would happen if the spindle fibers were disrupted during metaphase?
4. **a. Review** What is cytokinesis and when does it occur?
   b. Compare and Contrast How does cytokinesis differ in animal and plant cells?

WRITE ABOUT SCIENCE

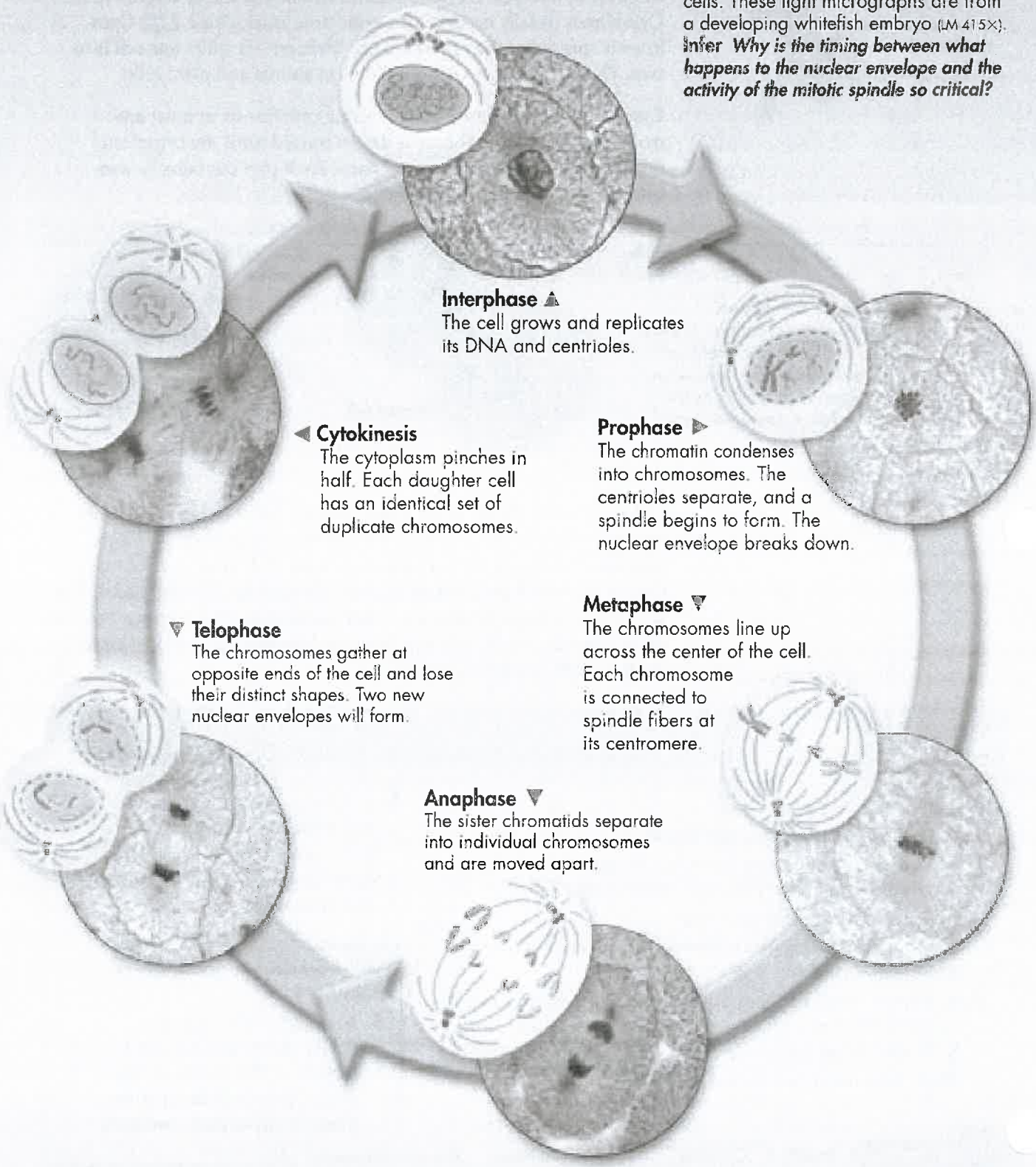
Summary

5. Summarize what happens during interphase. Be sure to include all three parts of interphase. *Hint:* Include all of the main details in your summary.

VISUAL SUMMARY

MITOSIS

FIGURE 10–13 The phases of mitosis shown here are typical of eukaryotic cells. These light micrographs are from a developing whitefish embryo (LM 415×). **Infer** ***Why is the timing between what happens to the nuclear envelope and the activity of the mitotic spindle so critical?***



Interphase
The cell grows and replicates its DNA and centrioles.

Prophase
The chromatin condenses into chromosomes. The centrioles separate, and a spindle begins to form. The nuclear envelope breaks down.

Metaphase
The chromosomes line up across the center of the cell. Each chromosome is connected to spindle fibers at its centromere.

Anaphase
The sister chromatids separate into individual chromosomes and are moved apart.

Telophase
The chromosomes gather at opposite ends of the cell and lose their distinct shapes. Two new nuclear envelopes will form.

Cytokinesis
The cytoplasm pinches in half. Each daughter cell has an identical set of duplicate chromosomes.

11.1 The Work of Gregor Mendel

Key Questions

Where does an organism get its unique characteristics?

How are different forms of a gene distributed to offspring?

Vocabulary

genetics • fertilization • trait • hybrid • gene • allele • principle of dominance • segregation • gamete

Taking Notes

Two-Column Chart Before you read, draw a line down the center of a sheet of paper. On the left side, write the main ideas in this lesson. On the right side, note the details and examples that support each of those ideas.

THINK ABOUT IT What is an inheritance? To many people, it is money or property left to them by relatives who have passed away. That kind of inheritance matters, of course, but there is another kind that matters even more. It is something we each receive from our parents—a contribution that determines our blood type, the color of our hair, and so much more. Most people leave their money and property behind by writing a will. But what kind of inheritance makes a person's face round or their hair curly?

The Experiments of Gregor Mendel

Where does an organism get its unique characteristics?

Every living thing—plant or animal, microbe or human being—has a set of characteristics inherited from its parent or parents. Since the beginning of recorded history, people have wanted to understand how that inheritance is passed from generation to generation. The delivery of characteristics from parent to offspring is called heredity. The scientific study of heredity, known as **genetics,** is the key to understanding what makes each organism unique.

The modern science of genetics was founded by an Austrian monk named Gregor Mendel. Mendel, shown in **Figure 11–1,** was born in 1822 in what is now the Czech Republic. After becoming a priest, Mendel spent several years studying science and mathematics at the University of Vienna. He spent the next 14 years working in a monastery and teaching high school. In addition to his teaching duties, Mendel was in charge of the monastery garden. In this simple garden, he was to do the work that changed biology forever.

Mendel carried out his work with ordinary garden peas, partly because peas are small and easy to grow. A single pea plant can produce hundreds of offspring. Today we call peas a "model system." Scientists use model systems because they are convenient to study and may tell us how other organisms, including humans, actually function. By using peas, Mendel was able to carry out, in just one or two growing seasons, experiments that would have been impossible to do with humans and that would have taken decades—if not centuries—to do with pigs, horses, or other large animals.

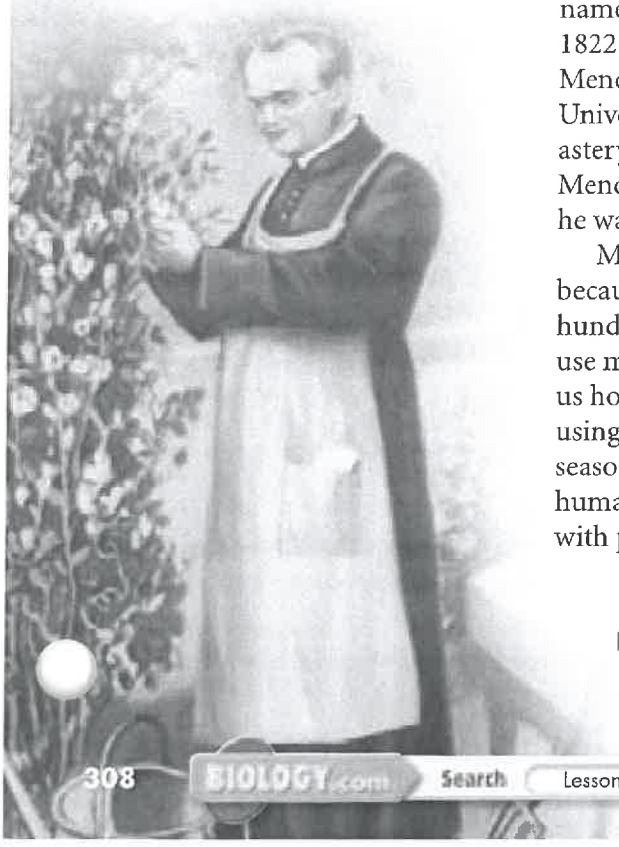


FIGURE 11–1 Gregor Mendel

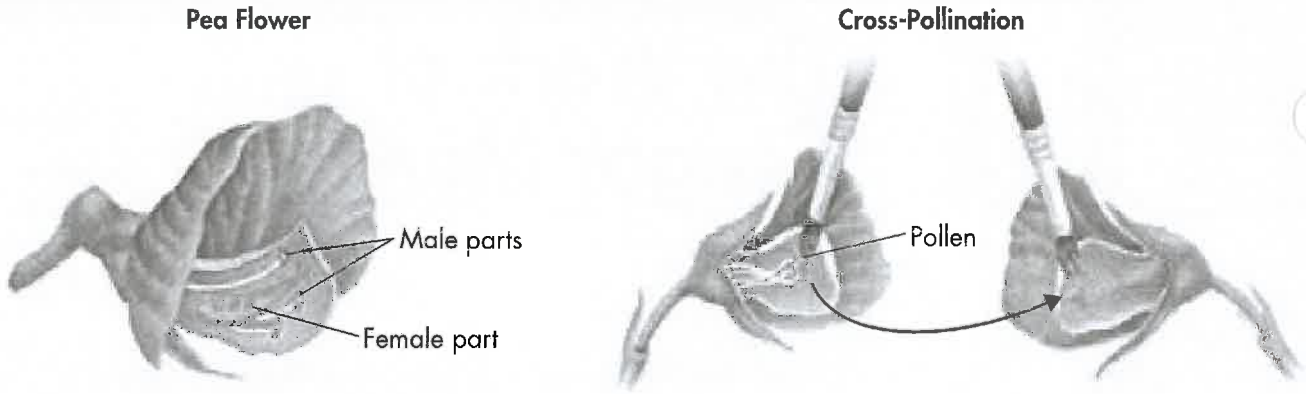


FIGURE 11–2 Cross-Pollination To cross-pollinate pea plants, Mendel cut off the male parts of one flower and then dusted the female part with pollen from another flower. **Apply Concepts** ***How did this procedure prevent self-pollination?***

The Role of Fertilization When Mendel began his experiments, he knew that the male part of each flower makes pollen, which contains the plant's male reproductive cells, called sperm. Similarly, Mendel knew that the female portion of each flower produces reproductive cells called eggs. During sexual reproduction, male and female reproductive cells join in a process known as **fertilization** to produce a new cell. In peas, this new cell develops into a tiny embryo encased within a seed.

Pea flowers are normally self-pollinating, which means that sperm cells fertilize egg cells from within the same flower. A plant grown from a seed produced by self-pollination inherits all of its characteristics from the single plant that bore it; it has a single parent.

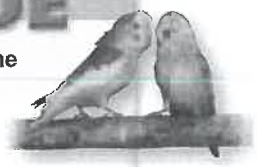
Mendel's monastery garden had several stocks of pea plants. These plants were "true-breeding," meaning that they were self-pollinating, and would produce offspring identical to themselves. In other words, the traits of each successive generation would be the same. A **trait** is a specific characteristic, such as seed color or plant height, of an individual. Many traits vary from one individual to another. For instance, one stock of Mendel's seeds produced only tall plants, while another produced only short ones. One line produced only green seeds, another produced only yellow seeds.

To learn how these traits were determined, Mendel decided to "cross" his stocks of true-breeding plants—that is, he caused one plant to reproduce with another plant. To do this, he had to prevent self-pollination. He did so by cutting away the pollen-bearing male parts of a flower. He then dusted the pollen from a different plant onto the female part of that flower, as shown in **Figure 11–2.** This process, known as cross-pollination, produces a plant that has two different parents. Cross-pollination allowed Mendel to breed plants with traits different from those of their parents and then study the results.

Mendel studied seven different traits of pea plants. Each of these seven traits had two contrasting characteristics, such as green seed color or yellow seed color. Mendel crossed plants with each of the seven contrasting characteristics and then studied their offspring. The offspring of crosses between parents with different traits are called **hybrids.**

In Your Notebook *Explain, in your own words, what fertilization is.*

MYSTERY CLUE



Parakeets come in four colors: white, green, blue, and yellow. How many alleles might there be for feather color?

Genes and Alleles When doing genetic crosses, we call each original pair of plants the P, or parental, generation. Their offspring are called the F_1, or first filial, generation. (*Filius* and *filia* are the Latin words for "son" and "daughter.")

What were Mendel's F_1 hybrid plants like? To his surprise, for each trait studied, all the offspring had the characteristics of only one of its parents, as shown in **Figure 11–3.** In each cross, the nature of the other parent, with regard to each trait, seemed to have disappeared. From these results, Mendel drew two conclusions. His first conclusion formed the basis of our current understanding of inheritance.

An individual's characteristics are determined by factors that are passed from one parental generation to the next. Today, scientists call the factors that are passed from parent to offspring **genes.**

Each of the traits Mendel studied was controlled by a single gene that occurred in two contrasting varieties. These variations produced different expressions, or forms, of each trait. For example, the gene for plant height occurred in one form that produced tall plants and in another form that produced short plants. The different forms of a gene are called **alleles** (uh LEELZ).

Dominant and Recessive Alleles Mendel's second conclusion is called the **principle of dominance.** This principle states that some alleles are dominant and others are recessive. An organism with at least one dominant allele for a particular form of a trait will exhibit that form of the trait. An organism with a recessive allele for a particular form of a trait will exhibit that form only when the dominant allele for the trait is not present. In Mendel's experiments, the allele for tall plants was dominant and the allele for short plants was recessive. Likewise, the allele for yellow seeds was dominant over the recessive allele for green seeds.

FIGURE 11–3 Mendel's F_1 Crosses When Mendel crossed plants with contrasting traits, the resulting hybrids had the traits of only one of the parents.

Mendel's Seven F_1 Crosses on Pea Plants

	Seed Shape	Seed Color	Seed Coat	Pod Shape	Pod Color	Flower Position	Plant Height
P	Round	Yellow	Gray	Smooth	Green	Axial	Tall
	X	X	X	X	X	X	X
	Wrinkled	Green	White	Constricted	Yellow	Terminal	Short
F_1	Round	Yellow	Gray	Smooth	Green	Axial	Tall

Quick Lab
GUIDED INQUIRY

Classroom Variation

1. Copy the data table into your notebook.
2. Write a prediction of whether the traits listed in the table will be evenly distributed or if there will be more dominant than recessive traits.
3. Examine your features, using a mirror if necessary. Determine which traits you have for features A–E.
4. Interview at least 14 other students to find out which traits they have. Tally the numbers. Record the totals in each column.

Trait Survey

Feature	Dominant Trait	Number	Recessive Trait	Number
A	Free ear lobes		Attached ear lobes	
B	Hair on fingers		No hair on fingers	
C	Widow's peak		No widow's peak	
D	Curly hair		Straight hair	
E	Cleft chin		Smooth chin	

Analyze and Conclude

1. **Calculate** Calculate the percentages of each trait in your total sample. How do these numbers compare to your prediction? MATH
2. **Form a Hypothesis** Why do you think recessive traits are more common in some cases?

In Your Notebook *Make a diagram that explains Mendel's principle of dominance.*

Segregation

How are different forms of a gene distributed to offspring?

Mendel didn't just stop after crossing the parent plants, because he had another question: Had the recessive alleles simply disappeared, or were they still present in the new plants? To find out, he allowed all seven kinds of F_1 hybrids to self-pollinate. The offspring of an F_1 cross are called the F_2 (second filial) generation. In effect, Mendel crossed the F_1 generation with itself to produce the F_2 offspring, as shown in **Figure 11–4.**

The F_1 Cross When Mendel compared the F_2 plants, he made a remarkable discovery: The traits controlled by the recessive alleles reappeared in the second generation. Roughly one fourth of the F_2 plants showed the trait controlled by the recessive allele. Why, then, did the recessive alleles seem to disappear in the F_1 generation, only to reappear in the F_2 generation?

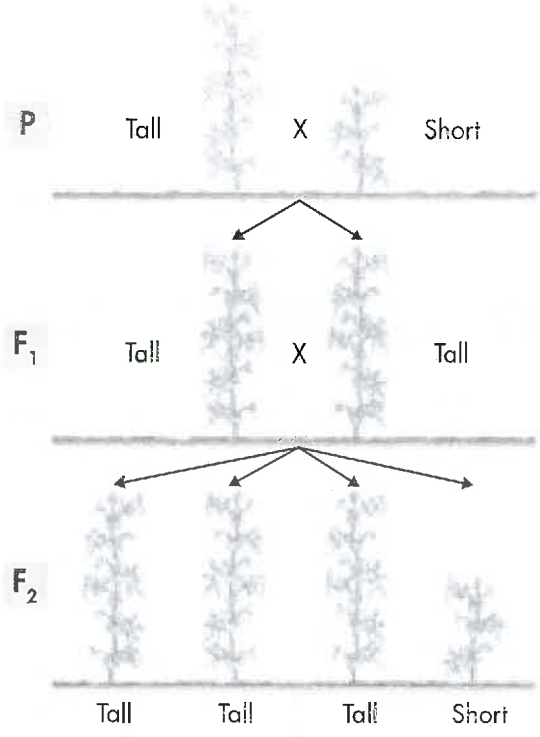


FIGURE 11-4 Results of the F_1 Cross When Mendel allowed the F_1 plants to reproduce by self-pollination, the traits controlled by recessive alleles reappeared in about one fourth of the F_2 plants in each cross. **Calculate** ***What proportion of the F_2 plants had a trait controlled by a dominant allele?*** MATH

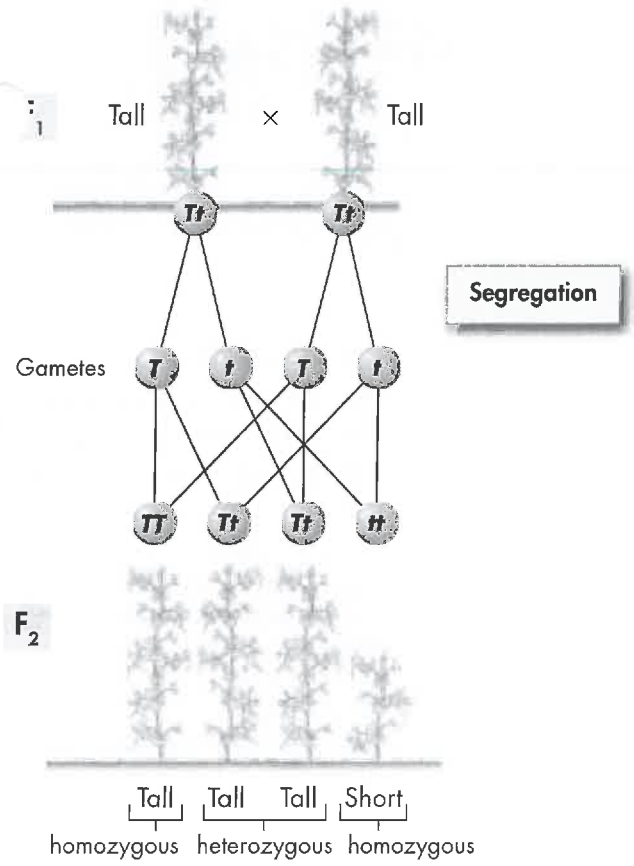


FIGURE 11–5 Segregation During gamete formation, alleles segregate from each other so that each gamete carries only a single copy of each gene. Each F_1 plant makes two types of gametes—those with the allele for tallness and those with the allele for shortness. The alleles are paired up again when gametes fuse during fertilization.

Explaining the F_1 Cross To begin with, Mendel assumed that a dominant allele had masked the corresponding recessive allele in the F_1 generation. However, the trait controlled by the recessive allele did show up in some of the F_2 plants. This reappearance indicated that, at some point, the allele for shortness had separated from the allele for tallness. How did this separation, or **segregation,** of alleles occur? Mendel suggested that the alleles for tallness and shortness in the F_1 plants must have segregated from each other during the formation of the sex cells, or **gametes** (GAM eetz). Did that suggestion make sense?

The Formation of Gametes Let's assume, as Mendel might have, that all the F_1 plants inherited an allele for tallness from the tall parent and one for shortness from the short parent. Because the allele for tallness is dominant, all the F_1 plants are tall. **During gamete formation, the alleles for each gene segregate from each other, so that each gamete carries only one allele for each gene.** Thus, each F_1 plant produces two kinds of gametes—those with the tall allele and those with the short allele.

Look at **Figure 11–5** to see how alleles separate during gamete formation and then pair up again in the F_2 generation. A capital letter represents a dominant allele. A lowercase letter represents a recessive allele. Now we can see why the recessive trait for height, *t*, reappeared in Mendel's F_2 generation. Each F_1 plant in Mendel's cross produced two kinds of gametes—those with the allele for tallness and those with the allele for shortness. Whenever a gamete that carried the *t* allele paired with the other gamete that carried the *t* allele to produce an F_2 plant, that plant was short. Every time one or both gametes of the pairing carried the *T* allele, a tall plant was produced. In other words, the F_2 generation had new combinations of alleles.

11.1 Assessment

Review Key Concepts

1. a. Review What did Mendel conclude determines biological inheritance?

b. Explain What are dominant and recessive alleles?

c. Apply Concepts Why were true-breeding pea plants important for Mendel's experiments?

2. a. Review What is segregation?

b. Explain What happens to alleles between the P generation and the F_2 generation?

c. Infer What evidence did Mendel use to explain how segregation occurs?

VISUAL THINKING

3. Use a diagram to explain Mendel's principles of dominance and segregation. Your diagram should show how alleles segregate during gamete formation.

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11.2 Applying Mendel's Principles

THINK ABOUT IT *Nothing in life is certain.* There's a great deal of wisdom in that old saying, and genetics is a fine example. If a parent carries two different alleles for a certain gene, we can't be sure which of those alleles will be inherited by any one of the parent's offspring. However, think carefully about the nature of inheritance and you'll see that even if we can't predict the exact future, we can do something almost as useful—we can figure out the odds.

Key Questions

- ***How can we use probability to predict traits?***
- ***How do alleles segregate when more than one gene is involved?***
- ***What did Mendel contribute to our understanding of genetics?***

Vocabulary

probability • homozygous • heterozygous • phenotype • genotype • Punnett square • independent assortment

Taking Notes

Preview Visuals Before you read, preview **Figure 11–7.** Try to infer the purpose of this diagram. As you read, compare your inference to the text. After you read, revise your statement if needed or write a new one about the diagram's purpose.

Probability and Punnett Squares

How can we use probability to predict traits?

Whenever Mendel performed a cross with pea plants, he carefully categorized and counted the offspring. Consequently, he had plenty of data to analyze. For example, whenever he crossed two plants that were hybrids for stem height (*Tt*), about three fourths of the resulting plants were tall and about one fourth were short.

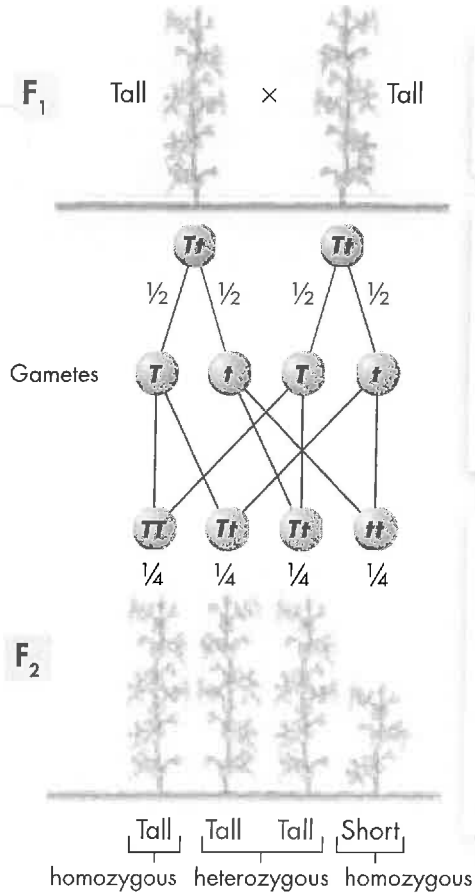
Upon analyzing his data, Mendel realized that the principles of probability could be used to explain the results of his genetic crosses. **Probability** is a concept you may have learned about in math class. It is the likelihood that a particular event will occur. As an example, consider an ordinary event, such as flipping a coin. There are two possible outcomes of this event: The coin may land either heads up or tails up. The chance, or probability, of either outcome is equal. Therefore, the probability that a single coin flip will land heads up is 1 chance in 2. This amounts to 1/2, or 50 percent.

If you flip a coin three times in a row, what is the probability that it will land heads up every time? Each coin flip is an independent event with a 1/2 probability of landing heads up. Therefore, the probability of flipping three heads in a row is:

$$1/2 \times 1/2 \times 1/2 = 1/8$$

As you can see, you have 1 chance in 8 of flipping heads three times in a row. The multiplication of individual probabilities illustrates an important point: Past outcomes do not affect future ones. Just because you've flipped three heads in a row does not mean that you're more likely to have a coin land tails up on the next flip. The probability for that flip is still 1/2.

FIGURE 11–6 Probability Probability allows you to calculate the likelihood that a particular event will occur. The probability that the coin will land heads up is ½, or 50 percent.



Both F_1 plants have the same set of alleles (*Tt*) and are tall.

The probability of each gamete acquiring the tall (*T*) allele is ½. Similarly, the probability of acquiring the short (*t*) allele is also ½.

When the alleles pair up in the F_2 generation, the probability of a tall offspring (*TT* or *Tt*) is ¼ + ¼ + ¼, or ¾. The probability that the offspring will be short (*tt*) is ¼.

FIGURE 11–7 Segregation and Probability In this cross, the *TT* and *Tt* allele combinations produced three tall pea plants, while the *tt* allele combination produced one short plant. These quantities follow the laws of probability. **Predict** ***If you crossed a* Tt *plant with a* Tt *plant, would the offspring be tall or short?***

Using Segregation to Predict Outcomes

The way in which alleles segregate during gamete formation is every bit as random as a coin flip. Therefore, the principles of probability can be used to predict the outcomes of genetic crosses.

Look again at Mendel's F_1 cross, shown in **Figure 11–7.** This cross produced a mixture of tall and short plants. Why were just 1/4 of the offspring short? Well, the F_1 plants were both tall. If each plant had one tall allele and one short allele (*Tt*), and if the alleles segregated as Mendel thought, then 1/2 of the gametes produced by the plants would carry the short allele (*t*). Yet, the *t* allele is recessive. The only way to produce a short (*tt*) plant is for two gametes, each carrying the *t* allele, to combine.

Like the coin toss, each F_2 gamete has a one in two, or 1/2, chance of carrying the *t* allele. There are two gametes, so the probability of both gametes carrying the *t* allele is $1/2 \times 1/2 = 1/4$. In other words, roughly one fourth of the F_2 offspring should be short, and the remaining three fourths should be tall. This predicted ratio—3 offspring exhibiting the dominant trait to 1 offspring exhibiting the recessive trait—showed up consistently in Mendel's experiments. For each of his seven crosses, about 3/4 of the plants showed the trait controlled by the dominant allele. About 1/4 showed the trait controlled by the recessive allele. Segregation did occur according to Mendel's model.

As you can see in the F_2 generation, not all organisms with the same characteristics have the same combinations of alleles. Both the *TT* and *Tt* allele combinations resulted in tall pea plants, but only one of these combinations contains identical alleles. Organisms that have two identical alleles for a particular gene—*TT* or *tt* in this example—are said to be **homozygous** (hoh moh zy gus). Organisms that have two different alleles for the same gene—such as *Tt*—are **heterozygous** (het ur oh zy gus).

Probabilities Predict Averages Probabilities predict the average outcome of a large number of events. If you flip a coin twice, you are likely to get one heads and one tails. However, you might also get two heads or two tails. To get the expected 50 : 50 ratio, you might have to flip the coin many times. The same is true of genetics.

The larger the number of offspring, the closer the results will be to the predicted values. If an F_2 generation contains just three or four offspring, it may not match Mendel's ratios. When an F_2 generation contains hundreds or thousands of individuals, the ratios usually come very close to matching predictions.

Genotype and Phenotype One of Mendel's most revolutionary insights followed directly from his observations of F_1 crosses: Every organism has a genetic makeup as well as a set of observable characteristics. All of the tall pea plants had the same **phenotype,** or physical traits. They did not, however, have the same **genotype,** or genetic makeup. Look again at **Figure 11–7** and you will find three different genotypes among the F_2 plants: *TT*, *Tt*, and *tt*. The genotype of an organism is inherited, and the phenotype is largely determined by the genotype. Two organisms may share the same phenotype but have different genotypes.

BUILD Vocabulary

PREFIXES The prefix *pheno-* in **phenotype** comes from the Greek word *phainein*, meaning "to show." *Geno-*, the prefix in **genotype,** is derived from the Greek word *genus*, meaning "race, kind."

Using Punnett Squares One of the best ways to predict the outcome of a genetic cross is by drawing a simple diagram known as a **Punnett square. Punnett squares use mathematical probability to help predict the genotype and phenotype combinations in genetic crosses.** Constructing a Punnett square is fairly easy. You begin with a square. Then, following the principle of segregation, all possible combinations of alleles in the gametes produced by one parent are written along the top edge of the square. The other parent's alleles are then segregated along the left edge. Next, every possible genotype is written into the boxes within the square, just as they might appear in the F_2 generation. **Figure 11–8** on the next page shows step-by-step instructions for constructing Punnett squares.

In Your Notebook *In your own words, write definitions for the terms* homozygous, heterozygous, phenotype, *and* genotype.

Quick Lab
GUIDED INQUIRY

How Are Dimples Inherited?

1. Write the last four digits of any telephone number. These four random digits represent the alleles of a gene that determines whether a person will have dimples. Odd digits represent the allele for the dominant trait of dimples. Even digits represent the allele for the recessive trait of no dimples.
2. Use the first two digits to represent a father's genotype. Use the symbols *D* and *d* to write his genotype as shown in the example.

Father's genotype is *dd* (2 even digits). Mother's genotype is *Dd* (1 even digit and 1 odd digit).

46 38

3. Use the last two digits the same way to find the mother's genotype. Write her genotype.
4. Use **Figure 11–8** on the next page to construct a Punnett square for the cross of these parents. Then, using the Punnett square, determine the probability that their child will have dimples.
5. Determine the class average of the percent of children with dimples.

Analyze and Conclude

1. **Apply Concepts** How does the class average compare with the result of a cross of two heterozygous parents?
2. **Draw Conclusions** What percentage of the children will be expected to have dimples if one parent is homozygous for dimples (*DD*) and the other is heterozygous (*Dd*)?

VISUAL SUMMARY

HOW TO MAKE A PUNNETT SQUARE

FIGURE 11–8 By drawing a Punnett square, you can determine the allele combinations that might result from a genetic cross.

One-Factor Cross

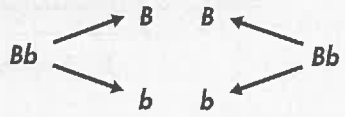
1 Start With the Parents

Write the genotypes of the two organisms that will serve as parents in a cross. In this example we will cross a male and female osprey, or fish hawk, that are heterozygous for large beaks. They each have genotypes of *Bb*.

Bb* and *Bb

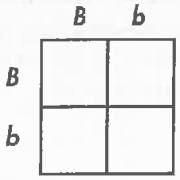
2 Figure Out the Gametes

Determine what alleles would be found in all of the possible gametes that each parent could produce.



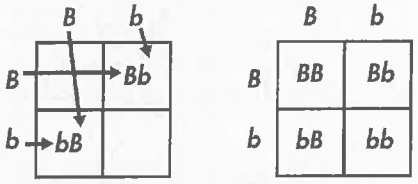
3 Line Them Up

Draw a table with enough squares for each pair of gametes from each parent. In this case, each parent can make two different types of gametes, *B* and *b*. Enter the genotypes of the gametes produced by both parents on the top and left sides of the table.



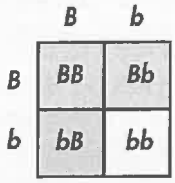
4 Write Out the New Genotypes

Fill in the table by combining the gametes' genotypes.



5 Figure Out the Results

Determine the genotype and phenotype of each offspring. Calculate the percentage of each. In this example, ¾ of the chicks will have large beaks, but only ½ will be heterozygous for this trait *(Bb)*.



Two-Factor Cross

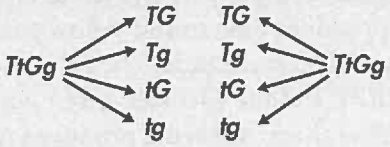
1 Start With the Parents

In this example we will cross two pea plants that are heterozygous for size (tall and short alleles) and pod color (green and yellow alleles). The genotypes of the two parents are *TtGg* and *TtGg*.

TtGg* and *TtGg

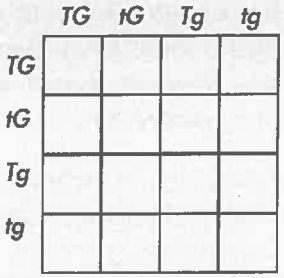
2 Figure Out the Gametes

Determine what alleles would be found in all of the possible gametes that each parent could produce.



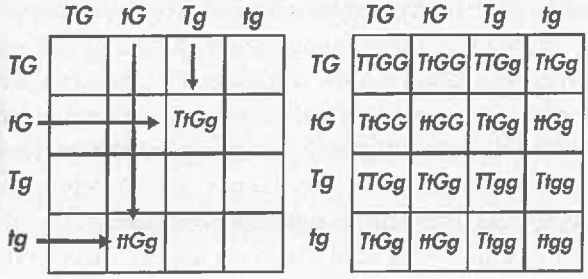
3 Line Them Up

In this case, each parent can make 4 different types of gametes, so the table needs to be 4 rows by 4 columns, or 16 squares.



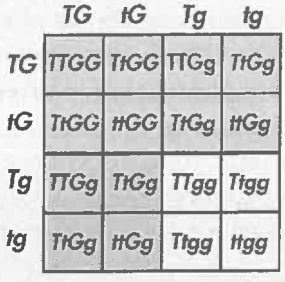
4 Write Out the New Genotypes

Fill in the table by combining the gametes' genotypes.



5 Figure Out the Results

In this example, the color of the squares represents pod color. Alleles written in black indicate short plants, while alleles written in red indicate tall plants.



Independent Assortment

How do alleles segregate when more than one gene is involved?

After showing that alleles segregate during the formation of gametes, Mendel wondered if the segregation of one pair of alleles affects another pair. For example, does the gene that determines the shape of a seed affect the gene for seed color? To find out, Mendel followed two different genes as they passed from one generation to the next. Because it involves two different genes, Mendel's experiment is known as a two-factor, or "dihybrid," cross. (Single-gene crosses are "monohybrid" crosses.)

The Two-Factor Cross: F_1 First, Mendel crossed true-breeding plants that produced only round yellow peas with plants that produced wrinkled green peas. The round yellow peas had the genotype *RRYY*, and the wrinkled green peas had the genotype *rryy*. All of the F_1 offspring produced round yellow peas. These results showed that the alleles for yellow and round peas are dominant. As the Punnett square in **Figure 11–9** shows, the genotype in each of these F_1 plants is *RrYy*. In other words, the F_1 plants were all heterozygous for both seed shape and seed color. This cross did not indicate whether genes assort, or segregate independently. However, it provided the hybrid plants needed to breed the F_2 generation.

The Two-Factor Cross: F_2 In the second part of this experiment, Mendel crossed the F_1 plants to produce F_2 offspring. Remember, each F_1 plant was formed by the fusion of a gamete carrying the dominant *RY* alleles with another gamete carrying the recessive *ry* alleles. Did this mean that the two dominant alleles would always stay together, or would they segregate independently, so that any combination of alleles was possible?

In Mendel's experiment, the F_2 plants produced 556 seeds. Mendel compared their variation. He observed that 315 of the seeds were round and yellow, while another 32 seeds were wrinkled and green—the two parental phenotypes. However, 209 seeds had combinations of phenotypes, and therefore combinations of alleles, that were not found in either parent. This clearly meant that the alleles for seed shape segregated independently of those for seed color. Put another way, genes that segregate independently (such as the genes for seed shape and seed color in pea plants) do not influence each other's inheritance.

Mendel's experimental results were very close to the 9 : 3 : 3 : 1 ratio that the Punnett square shown in **Figure 11–10** predicts. Mendel had discovered the principle of **independent assortment.** **The principle of independent assortment states that genes for different traits can segregate independently during the formation of gametes.** Independent assortment helps account for the many genetic variations observed in plants, animals, and other organisms—even when they have the same parents.

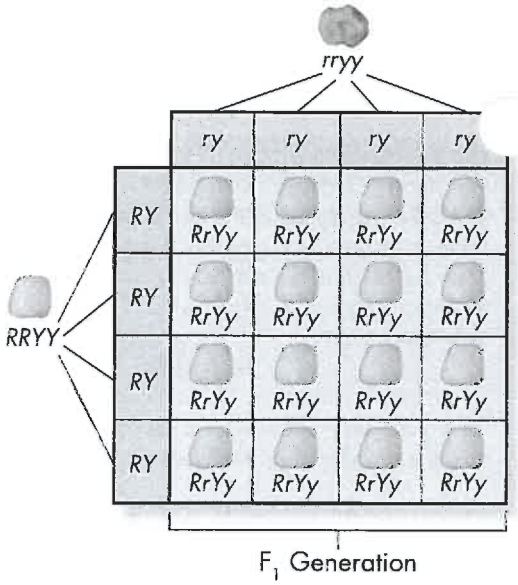


FIGURE 11–9 Two-Factor Cross: F_1 Mendel crossed plants that were homozygous dominant for round yellow peas with plants that were homozygous recessive for wrinkled green peas. All of the F_1 offspring were heterozygous dominant for round yellow peas. **Interpret Graphics** ***How is the genotype of the offspring different from that of the homozygous dominant parent?***

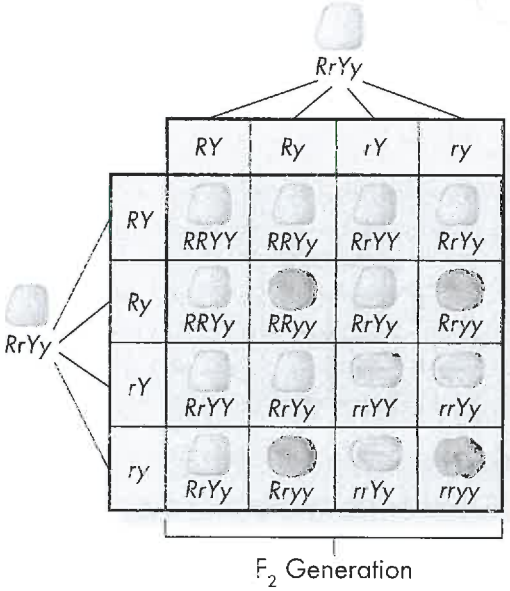


FIGURE 11–10 Two-Factor Cross: F_2 When Mendel crossed F_1 plants that were heterozygous dominant for round yellow peas, he found that the alleles segregated independently to produce the F_2 generation.

A Summary of Mendel's Principles

What did Mendel contribute to our understanding of genetics?

As you have seen, Mendel's principles of segregation and independent assortment can be observed through one- and two-factor crosses.

Mendel's principles of heredity, observed through patterns of inheritance, form the basis of modern genetics. These principles are as follows:

- The inheritance of biological characteristics is determined by individual units called genes, which are passed from parents to offspring.
- Where two or more forms (alleles) of the gene for a single trait exist, some alleles may be dominant and others may be recessive.
- In most sexually reproducing organisms, each adult has two copies of each gene—one from each parent. These genes segregate from each other when gametes are formed.
- Alleles for different genes usually segregate independently of each other.

Mendel's principles don't apply only to plants. At the beginning of the 1900s, the American geneticist Thomas Hunt Morgan wanted to use a model organism of another kind to advance the study of genetics. He decided to work on a tiny insect that kept showing up, uninvited, in his laboratory. The insect was the common fruit fly, *Drosophila melanogaster,* shown in **Figure 11–11.** *Drosophila* can produce plenty of offspring—a single pair can produce hundreds of young. Before long, Morgan and other biologists had tested all of Mendel's principles and learned that they applied to flies and other organisms as well. In fact, Mendel's basic principles can be used to study the inheritance of human traits and to calculate the probability of certain traits appearing in the next generation. You will learn more about human genetics in Chapter 14.

FIGURE 11–11 A Model Organism The common fruit fly, *Drosophila melanogaster,* is an ideal organism for genetic research. These fruit flies are poised on a lemon.

11.2 Assessment

Review Key Concepts

1. a. Review What is probability?

b. Use Models How are Punnett squares used to predict the outcomes of genetic crosses?

2. a. Review What is independent assortment?

b. Calculate An F_1 plant that is homozygous for shortness is crossed with a heterozygous F_1 plant. What is the probability that a seed from the cross will produce a tall plant? Use a Punnett square to explain your answer and to compare the probable genetic variations in the F_2 plants.

3. a. Review How did Gregor Mendel contribute to our understanding of inherited traits?

b. Apply Concepts Why is the fruit fly an ideal organism for genetic research?

Apply the Big idea

Information and Heredity

4. Suppose you are an avid gardener. One day, you come across a plant with beautiful lavender flowers. Knowing that the plant is self-pollinating, you harvest its seeds and plant them. Of the 106 plants that grow from these seeds, 31 have white flowers. Using a Punnett square, draw conclusions about the nature of the allele for lavender flowers.

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11.3 Other Patterns of Inheritance

THINK ABOUT IT Mendel's principles offer a tidy set of rules with which to predict various patterns of inheritance. Unfortunately, biology is not a tidy science. There are exceptions to every rule, and exceptions to the exceptions. What happens if one allele is not completely dominant over another? What if a gene has several alleles?

Key Questions

- ***What are some exceptions to Mendel's principles?***
- ***Does the environment have a role in how genes determine traits?***

Vocabulary

incomplete dominance • codominance • multiple allele • polygenic trait

Taking Notes

Outline Make an outline using the green and blue headings. As you read, write bulleted notes below each heading to summarize its topic.

Beyond Dominant and Recessive Alleles

What are some exceptions to Mendel's principles?

Despite the importance of Mendel's work, there are important exceptions to most of his principles. For example, not all genes show simple patterns of inheritance. In most organisms, genetics is more complicated, because the majority of genes have more than two alleles. Also, many important traits are controlled by more than one gene. Understanding these exceptions allows geneticists to predict the ways in which more complex traits are inherited.

Incomplete Dominance A cross between two four o'clock (*Mirabilis jalapa*) plants shows a common exception to Mendel's principles. **Some alleles are neither dominant nor recessive.** As shown in **Figure 11–12,** the F_1 generation produced by a cross between red-flowered (*RR*) and white-flowered (*WW*) *Mirabilis* plants consists of pink-colored flowers (*RW*). Which allele is dominant in this case? Neither one. Cases in which one allele is not completely dominant over another are called **incomplete dominance.** In incomplete dominance, the heterozygous phenotype lies somewhere between the two homozygous phenotypes.

Codominance A similar situation arises from **codominance,** in which the phenotypes produced by both alleles are clearly expressed. For example, in certain varieties of chicken, the allele for black feathers is codominant with the allele for white feathers. Heterozygous chickens have a color described as "erminette," speckled with black and white feathers. Unlike the blending of red and white colors in heterozygous four o'clocks, black and white colors appear separately in chickens. Many human genes, including one for a protein that controls cholesterol levels in the blood, show codominance, too. People with the heterozygous form of this gene produce two different forms of the protein, each with a different effect on cholesterol levels.

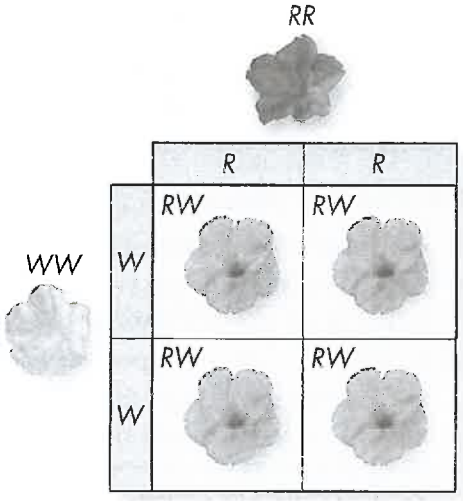


FIGURE 11–12 Incomplete Dominance In four o'clock plants, the alleles for red and white flowers show incomplete dominance. Heterozygous (*RW*) plants have pink flowers—a mix of red and white coloring.

Analyzing Data

Human Blood Types

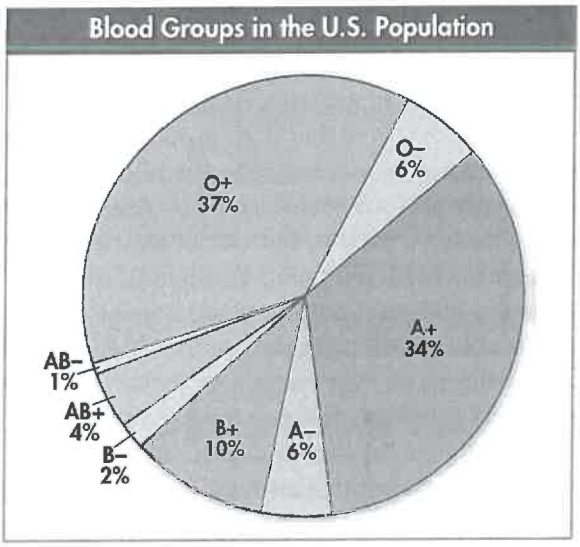
Red blood cells carry antigens, molecules that can trigger an immune reaction, on their surfaces. Human blood type A carries an A antigen, type B has a B antigen, type AB has both antigens, and type O carries neither antigen. The gene for these antigens has three alleles; A, B, and O.

For a transfusion to succeed, it must not introduce a new antigen into the body of the recipient. So, a person with type A blood may receive type O, but not vice versa.

Another gene controls a second type of antigen, known as Rh factor. Rh^+ individuals carry this antigen, while Rh^- ones don't. This chart of the U.S. population shows the percentage of each blood type.

1. Interpret Graphs Which blood type makes up the greatest percentage of the U.S. population?

2. Calculate What percentage of the total U.S. population has a positive Rh factor? What percentage has a negative Rh factor?



3. Infer Which blood type can be used for transfusion into the largest percentage of individuals? Which type has the smallest percentage of possible donors available?

4. Predict Could a person with O^+ blood have two parents with O^- blood? Could that person have a daughter with AB^+ blood? Explain your answers.

MYSTERY CLUE

Green feathers don't actually contain green pigments. Rather, they contain a mixture of blue and yellow pigments. Could feather color be controlled by more than one gene?

Multiple Alleles So far, our examples have described genes for which there are only two alleles, such as *a* and *A*. In nature, such genes are the exception rather than the rule. **Many genes exist in several different forms and are therefore said to have multiple alleles.** A gene with more than two alleles is said to have **multiple alleles.** An individual, of course, usually has only two copies of each gene, but many different alleles are often found within a population. One of the best-known examples is coat color in rabbits. A rabbit's coat color is determined by a single gene that has at least four different alleles. The four known alleles display a pattern of simple dominance that can produce four coat colors. Many other genes have multiple alleles, including the human genes for blood type.

Polygenic Traits **Many traits are produced by the interaction of several genes.** Traits controlled by two or more genes are said to be **polygenic traits.** *Polygenic* means "many genes." For example, at least three genes are involved in making the reddish-brown pigment in the eyes of fruit flies. Polygenic traits often show a wide range of phenotypes. The variety of skin color in humans comes about partly because more than four different genes probably control this trait.

In Your Notebook *In your own words, describe multiple alleles and polygenic traits. How are they similar? How are they different?*

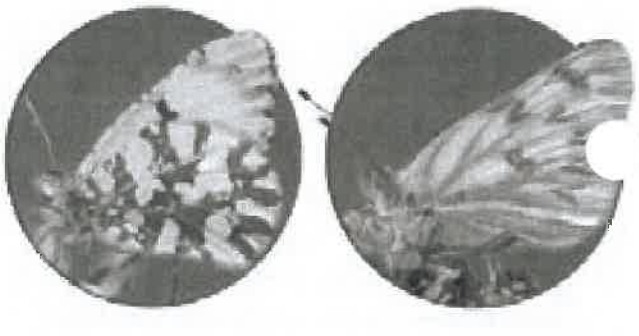
Genes and the Environment

Does the environment have a role in how genes determine traits?

The characteristics of any organism—whether plant, fruit fly, or human being—are not determined solely by the genes that organism inherits. Genes provide a plan for development, but how that plan unfolds also depends on the environment. In other words, the phenotype of an organism is only partly determined by its genotype.

Consider the western white butterfly, *Pontia occidentalis*, shown in **Figure 11–13.** It is found throughout western North America. Butterfly enthusiasts had noted for years that western whites hatching in the summer (right) had different color patterns on their wings than those hatching in the spring (left). Scientific studies showed the reason—butterflies hatching in the shorter days of springtime had greater levels of pigment in their wings, making their markings appear darker than those hatching in the longer days of summer. In other words, the environment in which the butterflies develop influences the expression of their genes for wing coloration. **Environmental conditions can affect gene expression and influence genetically determined traits.** An individual's actual phenotype is determined by its environment as well as its genes.

In the case of the western white butterfly, these changes in wing pigmentation are particularly important. In order to fly effectively, the body temperature of the butterfly must be 28°C–40°C (about 84°F–104°F). Since the spring months are cooler in the west, greater pigmentation helps them reach the body temperature needed for flight. Similarly, in the hot summer months, less pigmentation enables the moths to avoid overheating.



Environmental Temperature and Butterfly Needs		
Temp. Needed for Flight	Average Spring Temp.	Average Summer Temp.
28–40°C	26.5°C	34.8°C

FIGURE 11–13 Temperature and Wing Color Western white butterflies that hatch in the spring have darker wing patterns than those that hatch in summer. The dark wing color helps increase their body heat. This trait is important because the butterflies need to reach a certain temperature in order to fly. **Calculate** *What is the difference between the minimum temperature these butterflies need to fly and the average spring temperature? Would the same calculation apply to butterflies developing in the summer?* LOGICAL

11.3 Assessment

Review Key Concepts

1. **a. Review** What does *incomplete dominance* mean? Give an example.

   b. Design an Experiment Design an experiment to determine whether the pink flowers of petunia plants result from incomplete dominance.

2. **a. Review** What is the relationship between the environment and phenotype?

   b. Infer What might be the result of an exceptionally hot spring on wing pigmentation in the western white butterfly?

PRACTICE PROBLEM

3. Construct a genetics problem to be given as an assignment to a classmate. The problem must test incomplete dominance, codominance, multiple alleles, or polygenic traits. Your problem must have an answer key that includes all of your work.

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11.4 Meiosis

THINK ABOUT IT As geneticists in the early 1900s applied Mendel's principles, they wondered where genes might be located. They expected genes to be carried on structures inside the cell, but *which* structures? What cellular processes could account for segregation and independent assortment, as Mendel had described?

Chromosome Number

How many sets of genes are found in most adult organisms?

To hold true, Mendel's principles require at least two events to occur. First, an organism with two parents must inherit a single copy of every gene from each parent. Second, when that organism produces gametes, those two sets of genes must be separated so that each gamete contains just one set of genes. As it turns out, chromosomes—those strands of DNA and protein inside the cell nucleus—are the carriers of genes. The genes are located in specific positions on chromosomes.

Diploid Cells Consider the fruit fly that Morgan used, *Drosophila*. A body cell in an adult fruit fly has eight chromosomes, as shown in **Figure 11–14.** Four of the chromosomes come from its male parent, and four come from its female parent. These two sets of chromosomes are **homologous** (hoh MAHL uh gus), meaning that each of the four chromosomes from the male parent has a corresponding chromosome from the female parent. A cell that contains both sets of homologous chromosomes is said to be **diploid,** meaning "two sets." **The diploid cells of most adult organisms contain two complete sets of inherited chromosomes and two complete sets of genes.** The diploid number of chromosomes is sometimes represented by the symbol 2N. Thus, for *Drosophila,* the diploid number is 8, which can be written as $2N = 8$, where N represents the single set of chromosomes found in a sperm or egg cell.

Haploid Cells Some cells contain only a single set of chromosomes, and therefore a single set of genes. Such cells are **haploid,** meaning "one set." The gametes of sexually reproducing organisms, including fruit flies and peas, are haploid. For *Drosophila* gametes, the haploid number is 4, which can be written as $N = 4$.

Key Questions

- ***How many sets of genes are found in most adult organisms?***
- ***What events occur during each phase of meiosis?***
- ***How is meiosis different from mitosis?***
- ***How can two alleles from different genes be inherited together?***

Vocabulary

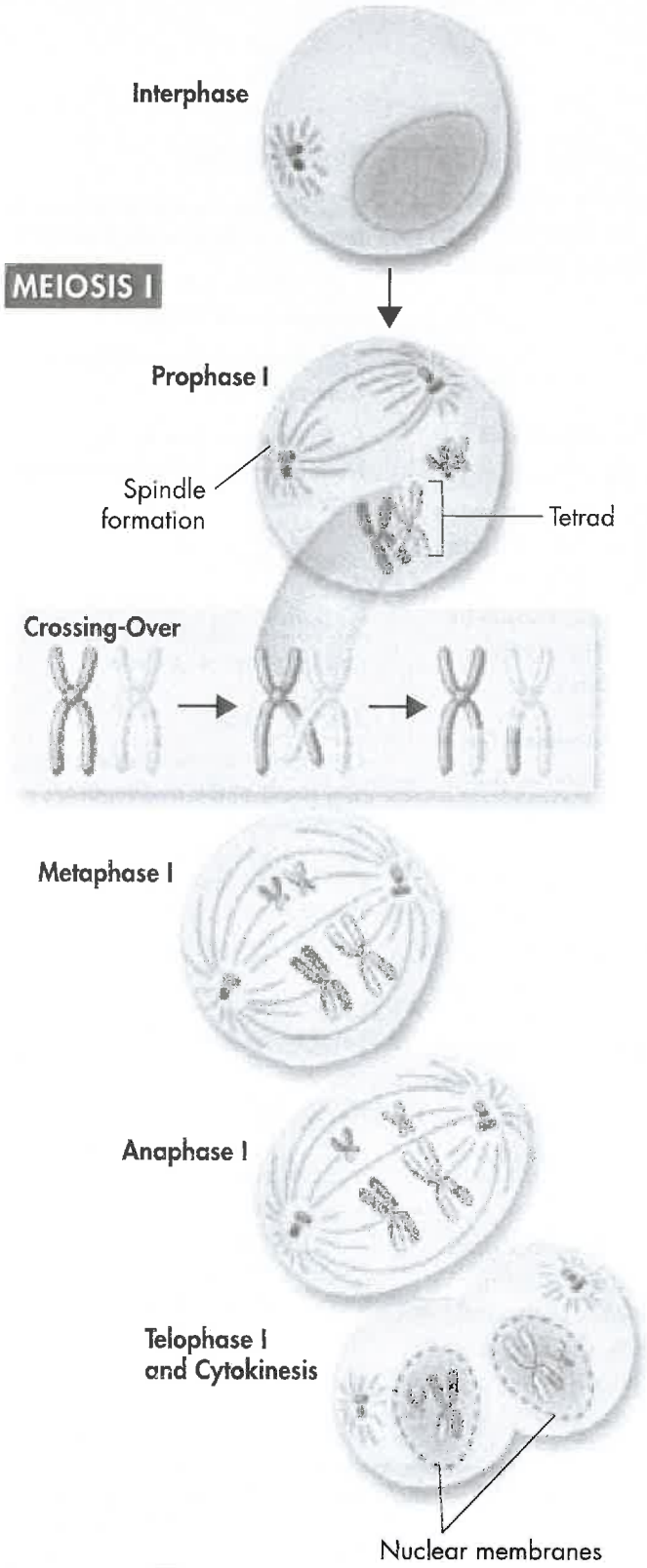
homologous • diploid • haploid • meiosis • tetrad • crossing-over • zygote

Taking Notes

Compare/Contrast Table Before you read, make a compare/contrast table to show the differences between mitosis and meiosis. As you read, complete the table.

FIGURE 11–14 Fruit Fly Chromosomes These chromosomes are from a fruit fly. Each of the fruit fly's body cells is diploid, containing eight chromosomes.

FIGURE 11–15 Meiosis I During meiosis I, a diploid cell undergoes a series of events that results in the production of two daughter cells. Neither daughter cell has the same sets of chromosomes that the original diploid cell had. **Interpret Graphics** ***How does crossing-over affect the alleles on a chromosome?***



Phases of Meiosis

What events occur during each phases of meiosis?

How are haploid (N) gamete cells produced from diploid (2N) cells? That's where meiosis (my OH sis) comes in. **Meiosis** is a process in which the number of chromosomes per cell is cut in half through the separation of homologous chromosomes in a diploid cell. Meiosis usually involves two distinct divisions, called meiosis I and meiosis II. By the end of meiosis II, the diploid cell becomes four haploid cells. Let's see how meiosis takes place in a cell that has a diploid number of 4 (2N = 4).

Meiosis I Just prior to meiosis I, the cell undergoes a round of chromosome replication during interphase. As in mitosis, which was discussed in Chapter 10, each replicated chromosome consists of two identical chromatids joined at the center. Follow the sequence in **Figure 11–15** as you read about meiosis I.

▶ ***Prophase I*** After interphase I, the cell begins to divide, and the chromosomes pair up. **In prophase I of meiosis, each replicated chromosome pairs with its corresponding homologous chromosome.** This pairing forms a structure called a **tetrad**, which contains four chromatids. As the homologous chromosomes form tetrads, they undergo a process called **crossing-over.** First, the chromatids of the homologous chromosomes cross over one another. Then, the crossed sections of the chromatids—which contain alleles—are exchanged. Crossing-over therefore produces new combinations of alleles in the cell.

▶ ***Metaphase I and Anaphase I*** As prophase I ends, a spindle forms and attaches to each tetrad. **During metaphase I of meiosis, paired homologous chromosomes line up across the center of the cell.** As the cell moves into anaphase I, the homologous pairs of chromosomes separate. **During anaphase I, spindle fibers pull each homologous chromosome pair toward opposite ends of the cell.**

▶ ***Telophase I and Cytokinesis*** When anaphase I is complete, the separated chromosomes cluster at opposite ends of the cell. **The next phase is telophase I, in which a nuclear membrane forms around each cluster of chromosomes. Cytokinesis follows telophase I, forming two new cells.**

Meiosis I results in two cells, called daughter cells. However, because each pair of homologous chromosomes was separated, neither daughter cell has the two complete sets of chromosomes that it would have in a diploid cell. Those two sets have been shuffled and sorted almost like a deck of cards. The two cells produced by meiosis I have sets of chromosomes and alleles that are different from each other and from the diploid cell that entered meiosis I.

Meiosis II The two cells now enter a second meiotic division. Unlike the first division, neither cell goes through a round of chromosome replication before entering meiosis II.

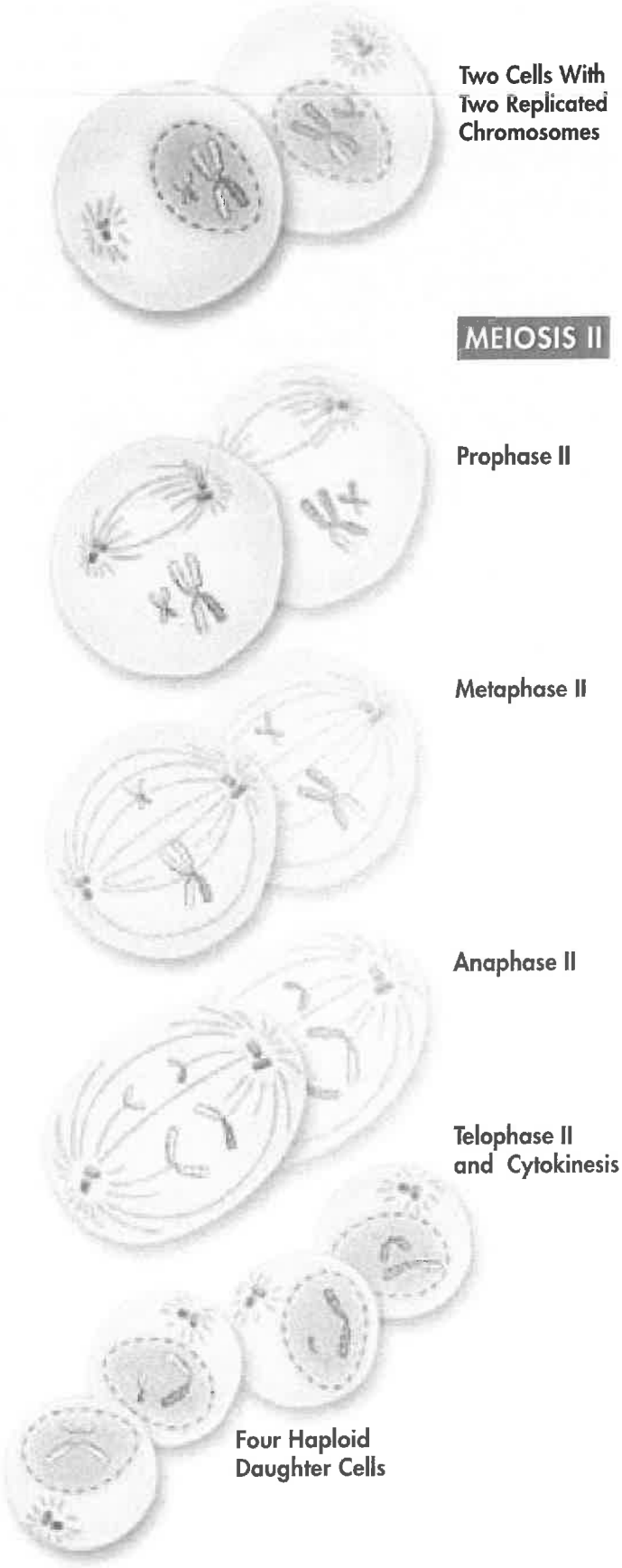
▶ ***Prophase II*** **As the cells enter prophase II, their chromosomes—each consisting of two chromatids—become visible.** The chromosomes do not pair to form tetrads, because the homologous pairs were already separated during meiosis I.

▶ ***Metaphase II, Anaphase II, Telophase II, and Cytokinesis*** During metaphase of meiosis II, chromosomes line up in the center of each cell. As the cell enters anaphase, the paired chromatids separate. **The final four phases of meiosis II are similar to those in meiosis I. However, the result is four haploid daughter cells.** In the example shown here, each of the four daughter cells produced in meiosis II receive two chromosomes. These four daughter cells now contain the haploid number (N)—just two chromosomes each.

Gametes to Zygotes The haploid cells produced by meiosis II are the gametes that are so important to heredity. In male animals, these gametes are called sperm. In some plants, pollen grains contain haploid sperm cells. In female animals, generally only one of the cells produced by meiosis is involved in reproduction. The female gamete is called an egg in animals and an egg cell in some plants. After it is fertilized, the egg is called a **zygote** (zy goht). The zygote undergoes cell division by mitosis and eventually forms a new organism.

In Your Notebook *Describe the difference between meiosis I and meiosis II. How are the end results different?*

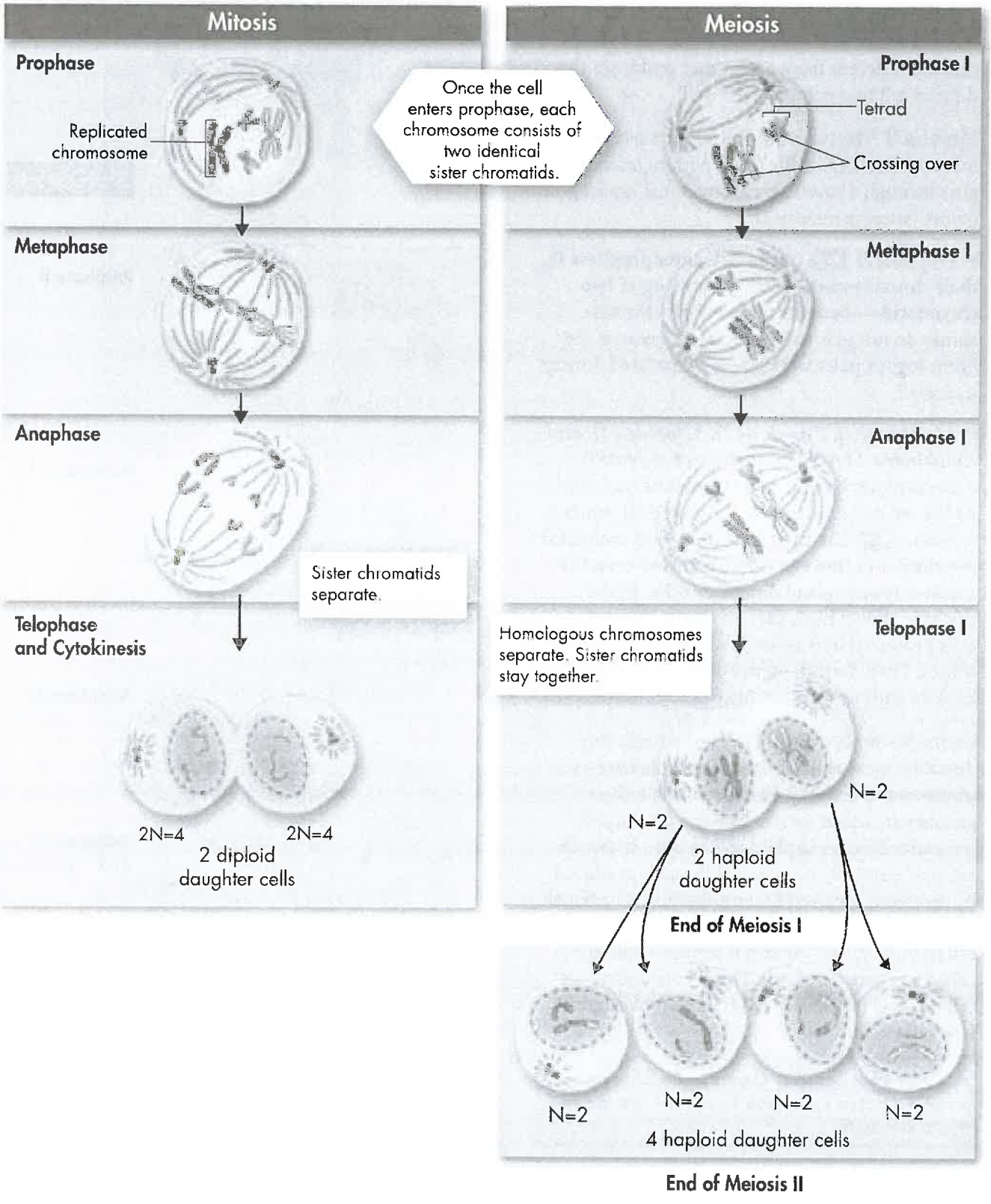
FIGURE 11–16 Meiosis II The second meiotic division, called meiosis II, produces four haploid daughter cells.



VISUAL SUMMARY

COMPARING MITOSIS AND MEIOSIS

FIGURE 11–17 Mitosis and meiosis both ensure that cells inherit genetic information. Both processes begin after interphase, when chromosome replication occurs. However, the two processes differ in the separation of chromosomes, the number of cells produced, and the number of chromosomes each cell contains.



Comparing Meiosis and Mitosis

How is meiosis different from mitosis?

The words *mitosis* and *meiosis* may sound similar, but the two processes are very different, as you can see in **Figure 11–17.** Mitosis can be a form of asexual reproduction, whereas meiosis is an early step in sexual reproduction. There are three other ways in which these two processes differ.

Replication and Separation of Genetic Material Mitosis and meiosis are both preceded by a complete copying, or replication, of the genetic material of chromosomes. However, the next steps differ dramatically. **In mitosis, when the two sets of genetic material separate, each daughter cell receives one complete set of chromosomes. In meiosis, homologous chromosomes line up and then move to separate daughter cells.** As a result, the two alleles for each gene are segregated, and end up in different cells. The sorting and recombination of genes in meiosis result in a greater variety of possible gene combinations than could result from mitosis.

Changes in Chromosome Number **Mitosis does not normally change the chromosome number of the original cell. This is not the case for meiosis, which reduces the chromosome number by half.** A diploid cell that enters mitosis with eight chromosomes will divide to produce two diploid daughter cells, each of which also has eight chromosomes. On the other hand, a diploid cell that enters meiosis with eight chromosomes will pass through two meiotic divisions to produce four haploid gamete cells, each with only four chromosomes.

Analyzing Data

Calculating Haploid and Diploid Numbers

Haploid and diploid numbers are designated by the algebraic notations N and 2N, respectively. Either number can be calculated when the other is known. For example, if the haploid number (N) is 3, the diploid number (2N) is 2×3, or 6. If the diploid number (2N) is 12, the haploid number (N) is 12/2, or 6.

The table shows haploid or diploid numbers of a variety of organisms. Copy the table into your notebook and complete it. Then, use the table to answer the questions that follow.

Trait Survey

Organism	Haploid Number	Diploid Number
Amoeba	N=25	
Chimpanzee	N=24	
Earthworm	N=18	
Fern		2N=1010
Hamster	N=22	
Human		2N=46
Onion		2N=16

1. **Calculate** What are the haploid numbers for the fern and onion plants? MATH
2. **Interpret Data** In the table, which organisms' diploid numbers are closest to that of a human?
3. **Apply Concepts** Why is a diploid number always even?
4. **Evaluate** Which organism's haploid and diploid numbers do you find the most surprising? Why?

Number of Cell Divisions Mitosis is a single cell division, resulting in the production of two identical daughter cells. On the other hand, meiosis requires two rounds of cell division, and, in most organisms, produces a total of four daughter cells. **Mitosis results in the production of two genetically identical diploid cells, whereas meiosis produces four genetically different haploid cells.**

Gene Linkage and Gene Maps

How can two alleles from different genes be inherited together?

If you think carefully about Mendel's principle of independent assortment in relation to meiosis, one question might bother you. Genes that are located on different chromosomes assort independently, but what about genes that are located on the same chromosome? Wouldn't they generally be inherited together?

Gene Linkage The answer to this question, as Thomas Hunt Morgan first realized in 1910, is yes. Morgan's research on fruit flies led him to the principle of gene linkage. After identifying more than 50 *Drosophila* genes, Morgan discovered that many of them appeared to be "linked" together in ways that, at first glance, seemed to violate the principle of independent assortment. For example, Morgan used a fly with reddish-orange eyes and miniature wings in a series of test crosses. His results showed that the genes for those two traits were almost always inherited together. Only rarely did the genes separate from each other. Morgan and his associates observed so many genes that were inherited together that, before long, they could group all of the fly's genes into four linkage groups. The linkage groups assorted independently, but all of the genes in one group were inherited together. As it turns out, *Drosophila* has four linkage groups and four pairs of chromosomes.

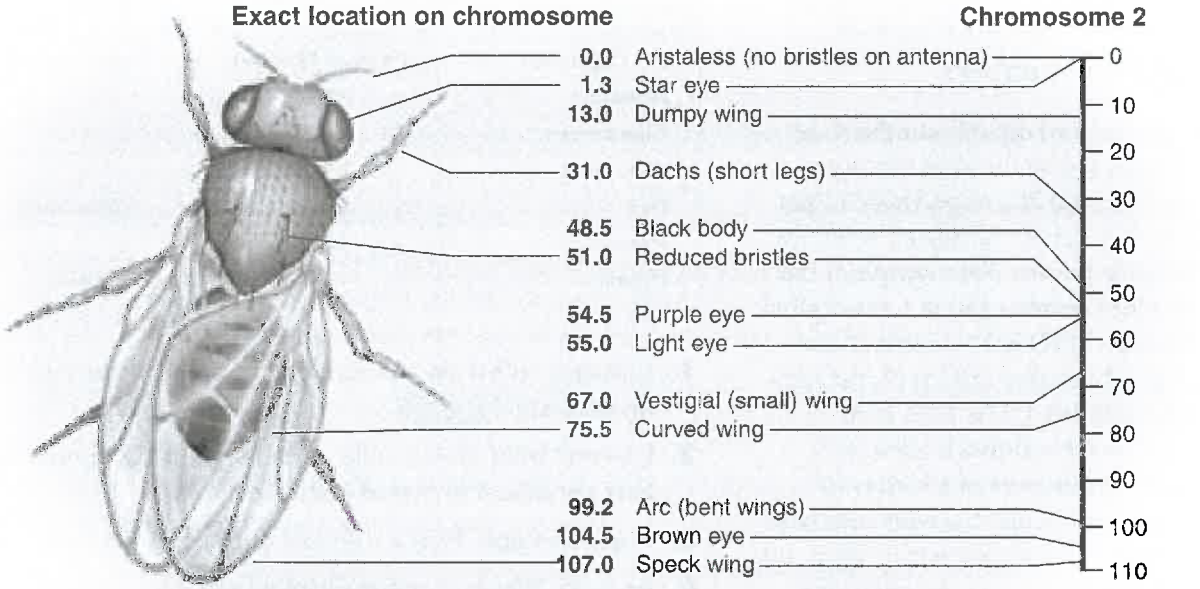


FIGURE 11–18 Gene Map This gene map shows the location of a variety of genes on chromosome 2 of the fruit fly. The genes are named after the problems that abnormal alleles cause, *not* after the normal structures. **Interpret Graphics** ***Where on the chromosome is the "purple eye" gene located?***

Morgan's findings led to two remarkable conclusions. First, each chromosome is actually a group of linked genes. Second, Mendel's principle of independent assortment still holds true. It is the chromosomes, however, that assort independently, not individual genes. **Alleles of different genes tend to be inherited together from one generation to the next when those genes are located on the same chromosome.**

How did Mendel manage to miss gene linkage? By luck, or design, several of the genes he studied are on different chromosomes. Others are so far apart that they also assort independently.

MYSTERY CLUE

White is the least common color found in parakeets. What does this fact suggest about the genotypes of both green parents?

Gene Mapping In 1911, a Columbia University student was working part time in Morgan's lab. This student, Alfred Sturtevant, wondered if the frequency of crossing-over between genes during meiosis might be a clue to the genes' locations. Sturtevant reasoned that the farther apart two genes were on a chromosome, the more likely it would be that crossing-over would occur between them. If two genes are close together, then crossovers between them should be rare. If two genes are far apart, then crossovers between them should be more common. By this reasoning, he could use the frequency of crossing-over between genes to determine their distances from each other.

Sturtevant gathered up several notebooks of lab data and took them back to his room. The next morning, he presented Morgan with a gene map showing the relative locations of each known gene on one of the *Drosophila* chromosomes. Sturtevant's method has been used to construct gene maps, like the one in **Figure 11–18,** ever since this discovery.

11.4 Assessment

Review Key Concepts

1. a. Review Describe the main results of meiosis.

b. Calculate In human cells, 2N = 46. How many chromosomes would you expect to find in a sperm cell? How many would you expect to find in an egg cell?

2. a. Review Write a summary of each phase of meiosis.

b. Use Analogies Compare the chromosomes of a diploid cell to a collection of shoes in a closet. How are they similar? What would make the shoe collection comparable to the chromosomes of a haploid cell?

3. a. Review What are the principal differences between mitosis and meiosis?

b. Apply Concepts Is there any difference between sister chromatids and homologous pairs of chromosomes? Explain.

4. a. Review How does the principle of independent assortment apply to chromosomes?

b. Infer If two genes are on the same chromosome but usually assort independently, what does that tell you about how close together they are?

Apply the Big idea

Information and Heredity

5. In asexual reproduction, mitosis occurs but meiosis does not occur. Which type of reproduction—sexual or asexual—results in offspring with greater genetic variation? Explain your answer.

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14.1 Human Chromosomes

Key Questions

- ***What is a karyotype?***
- ***What patterns of inheritance do human traits follow?***
- ***How can pedigrees be used to analyze human inheritance?***

Vocabulary

genome • karyotype • sex chromosome • autosome • sex-linked gene • pedigree

Taking Notes

Outline Before you read, make an outline of the major headings in the lesson. As you read, fill in main ideas and supporting details for each heading.

THINK ABOUT IT If you had to pick an ideal organism for the study of genetics, would you choose one that produced lots of offspring? How about one that was easy to grow in the lab? Would you select one with a short life span in order to do several crosses per month? How about all of the above? You certainly would not choose an organism that produced very few offspring, had a long life span, and could not be grown in a lab. Yet, when we study human genetics, this is exactly the sort of organism we deal with. Given all of these difficulties, it may seem a wonder that we know as much about human genetics as we do.

Karyotypes

What is a karyotype?

What makes us human? We might try to answer that question by looking under the microscope to see what is inside a human cell. Not surprisingly, human cells look much like the cells of other animals. To find what makes us uniquely human, we have to look deeper, into the genetic instructions that build each new individual. To begin this undertaking, we have to explore the human genome. A **genome** is the full set of genetic information that an organism carries in its DNA.

The study of any genome starts with chromosomes—those bundles of DNA and protein found in the nuclei of eukaryotic cells. To see human chromosomes clearly, cell biologists photograph cells in mitosis, when the chromosomes are fully condensed and easy to view. Scientists then cut out the chromosomes from the photographs and arrange them in a picture known as a **karyotype** (KAR ee uh typ). **A karyotype shows the complete diploid set of chromosomes grouped together in pairs, arranged in order of decreasing size.**

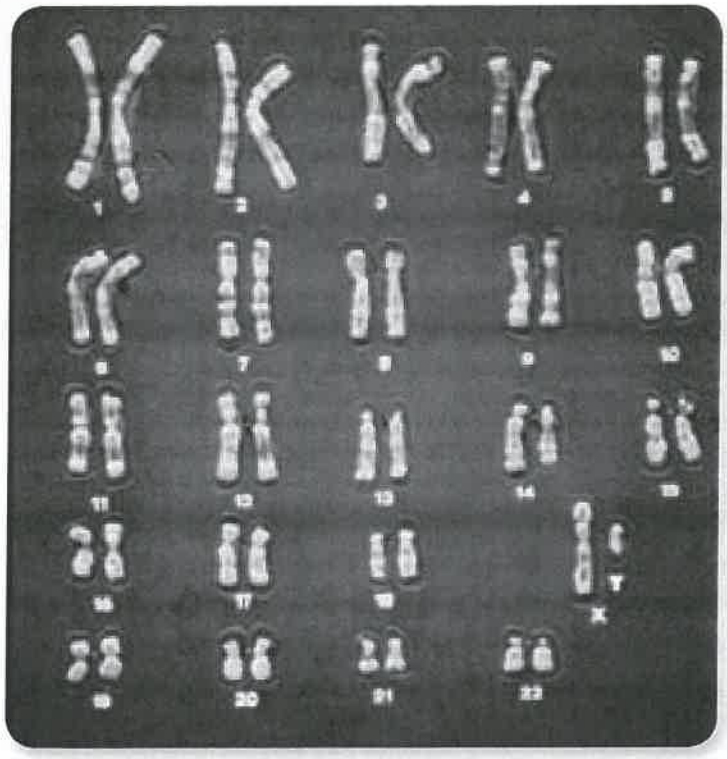

FIGURE 14–1 A Human Karyotype A typical human cell has 23 pairs of chromosomes. These chromosomes have been cut out of a photograph and arranged to form a karyotype.

The karyotype in **Figure 14–1** is from a typical human cell, which contains 46 chromosomes, arranged in 23 pairs. Why do our chromosomes come in pairs? Remember that we begin life when a haploid sperm, carrying just 23 chromosomes, fertilizes a haploid egg, also with 23 chromosomes. The resulting diploid cell develops into a new individual and carries the full complement of 46 chromosomes—two sets of 23.

Sex Chromosomes Two of the 46 chromosomes in the human genome are known as **sex chromosomes,** because they determine an individual's sex. Females have two copies of the X chromosome. Males have one X chromosome and one Y chromosome. As you can see in **Figure 14–2,** this is the reason why males and females are born in a roughly 50 : 50 ratio. All human egg cells carry a single X chromosome (23,X). However, half of all sperm cells carry an X chromosome (23,X) and half carry a Y chromosome (23,Y). This ensures that just about half the zygotes will be males and half will be females.

More than 1200 genes are found on the X chromosome, some of which are shown in **Figure 14–3.** Note that the human Y chromosome is much smaller than the X chromosome and contains only about 140 genes, most of which are associated with male sex determination and sperm development.

Autosomal Chromosomes To distinguish them from the sex chromosomes, the remaining 44 human chromosomes are known as autosomal chromosomes, or **autosomes.** The complete human genome consists of 46 chromosomes, including 44 autosomes and 2 sex chromosomes. To quickly summarize the total number of chromosomes present in a human cell—both autosomes and sex chromosomes—biologists write 46,XX for females and 46,XY for males.

In Your Notebook *Describe what makes up a human karyotype.*

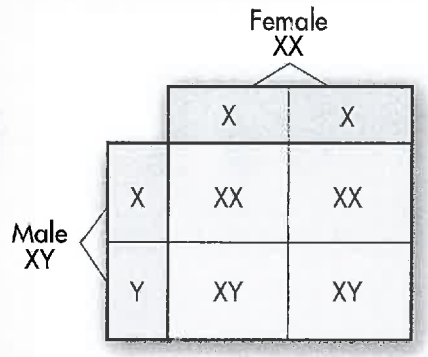

FIGURE 14–2 Sex Ratios Human egg cells contain a single X chromosome. Sperm cells contain either one X chromosome or one Y chromosome. **Interpret Tables** ***What does this Punnett square suggest about the sex ratio of the human population?***

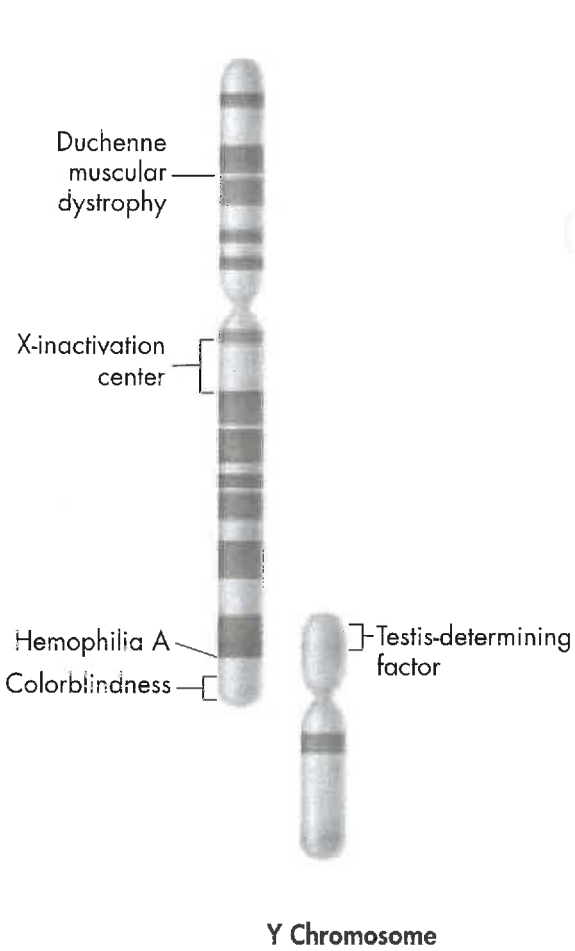

FIGURE 14–3 X and Y Chromosomes The human Y chromosome is smaller and carries fewer genes than the human X chromosome.

FIGURE 14–4 Recessive Alleles Some of the recessive alleles of the *MC1R* gene cause red hair. An individual with red hair usually has two of these recessive alleles.

Transmission of Human Traits

What patterns of inheritance do human traits follow?

It has not been easy studying our species using traditional genetic techniques. Despite the difficulties, human genetics has progressed rapidly, especially in recent years, with the use of molecular techniques to study human DNA. What have these studies shown? Human genes follow the same Mendelian patterns of inheritance as the genes of other organisms.

Dominant and Recessive Alleles

Many human traits follow a pattern of simple dominance. For instance, a gene known as *MC1R* helps determine skin and hair color. Some of *MC1R*'s recessive alleles produce red hair. An individual with red hair usually has two of these recessive alleles, inheriting a copy from each parent. Dominant alleles for the *MC1R* gene help produce darker hair colors.

Another trait that displays simple dominance is the Rhesus, or Rh blood group. The allele for Rh factor comes in two forms: Rh^+ and Rh^-. Rh^+ is dominant, so an individual with both alleles (Rh^+/Rh^-) is said to have Rh positive blood. Rh negative blood is found in individuals with two recessive alleles (Rh^-/Rh^-).

Codominant and Multiple Alleles

The alleles for many human genes display codominant inheritance. One example is the ABO blood group, determined by a gene with three alleles: I^A, I^B, and i. Alleles I^A and I^B are codominant. They produce molecules known as antigens on the surface of red blood cells. As **Figure 14–5** shows, individuals with alleles I^A and I^B produce both A and B antigens, making them blood type AB. The i allele is recessive. Individuals with alleles I^AI^A or I^Ai produce only the A antigen, making them blood type A. Those with I^BI^B or I^Bi alleles are type B. Those homozygous for the i allele (ii) produce no antigen and are said to have blood type O. If a patient has AB-negative blood, it means the individual has I^A and I^B alleles from the ABO gene and two Rh^- alleles from the Rh gene.

FIGURE 14–5 Human Blood Groups This table shows the relationship between genotype and phenotype for the ABO blood group. It also shows which blood types can safely be transfused into people with other blood types. **Apply Concepts** ***How can there be four different phenotypes even though there are six different genotypes?***

Blood Groups

Phenotype (Blood Type)	Genotype	Antigen on Red Blood Cell	Safe Transfusions	
			To	From
A	I^AI^A or I^Ai	A	A, AB	A, O
B	I^BI^B or I^Bi	B	B, AB	B, O
AB	I^AI^B	A and B	AB	A, B, AB, O
O	ii	None	A, B, AB, O	O

Sex-Linked Inheritance **Because the X and Y chromosomes determine sex, the genes located on them show a pattern of inheritance called sex-linkage.** A **sex-linked gene** is a gene located on a sex chromosome. As you might expect, genes on the Y chromosome are found only in males and are passed directly from father to son. Genes located on the X chromosome are found in both sexes, but the fact that men have just one X chromosome leads to some interesting consequences.

For example, humans have three genes responsible for color vision, all located on the X chromosome. In males, a defective allele for any of these genes results in colorblindness, an inability to distinguish certain colors. The most common form, red-green colorblindness, occurs in about 1 in 12 males. Among females, however, colorblindness affects only about 1 in 200. Why is there such a difference? In order for a recessive allele, like colorblindness, to be expressed in females, it must be present in two copies—one on each of the X chromosomes. This means that the recessive phenotype of a sex-linked genetic disorder tends to be much more common among males than among females.

MYSTERY CLUE

The presence of two sickle cell alleles is needed to produce sickle cell disease. Males and females develop sickle cell disease in equal frequencies. What do these statements suggest about the location of the gene responsible for the disorder?

Quick Lab
GUIDED INQUIRY

How Is Colorblindness Transmitted?

1. Make a data table with the column headings Trial, Colors, Sex of Individual, and Number of X-Linked Alleles. Draw ten rows under the headings and fill in the numbers 1 through 10 in the Trial column. Label one plastic cup Mother and a second plastic cup Father.

2. The white beans represent X chromosomes. Use a black marker to make a dot on 1 white bean to represent the X-linked allele for colorblindness. Place this bean, plus 1 unmarked white bean, into the cup labeled Mother.

3. Mark a black dot on 1 more white bean. Place this bean, plus 1 red bean, into the cup labeled Father. The red bean represents a Y chromosome.

4. Close your eyes and pick one bean from each cup to represent how each parent contributes to a sex chromosome and a fertilized egg.

5. In your data table, record the color of each bean and the sex of an individual who would carry this pair of sex chromosomes. Also record how many X-linked alleles the individual has. Put the beans back in the cups they came from.

6. Determine whether the individual would have colorblindness.

7. Repeat steps 4 to 6 for a total of 10 pairs of beans.

Analyze and Conclude

1. Draw Conclusions How do human sex chromosomes keep the numbers of males and females roughly equal?

2. Calculate Calculate the class totals for each data column. How many females were colorblind? How many males? Explain these results.

3. Use Models Evaluate your model. How accurately does it represent the transmission of colorblindness in a population? Why?

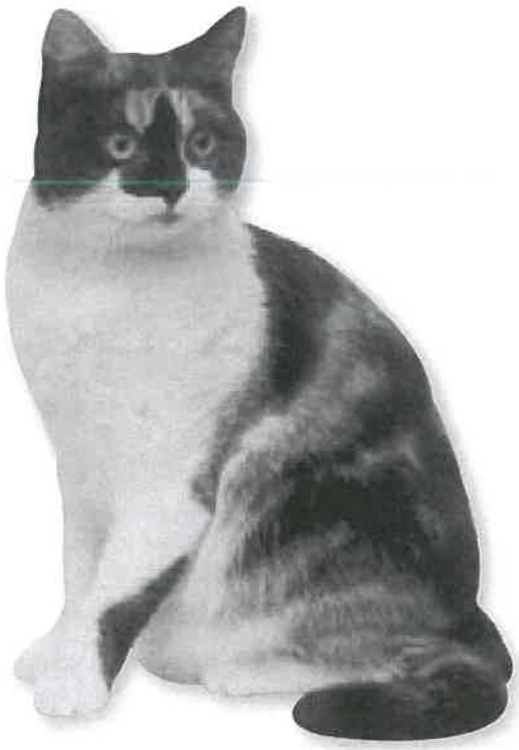

FIGURE 14–6 X-Chromosome Inactivation Female calico cats are tri-colored. The color of spots on their fur is controlled by a gene on the X chromosome. Spots are either orange or black, depending on which X chromosome is inactivated in different patches of their skin.

X-Chromosome Inactivation If just one X chromosome is enough for cells in males, how does the cell "adjust" to the extra X chromosome in female cells? The answer was discovered by the British geneticist Mary Lyon. In female cells, most of the genes in one of the X chromosomes are randomly switched off, forming a dense region in the nucleus known as a Barr body. Barr bodies are generally not found in males because their single X chromosome is still active.

The same process happens in other mammals. In cats, for example, a gene that controls the color of coat spots is located on the X chromosome. One X chromosome may have an allele for orange spots and the other X chromosome may have an allele for black spots. In cells in some parts of the body, one X chromosome is switched off. In other parts of the body, the other X chromosome is switched off. As a result, the cat's fur has a mixture of orange and black spots, like those in **Figure 14–6.** Male cats, which have just one X chromosome, can have spots of only one color. Therefore, if the cat's fur has three colors—white with orange and black spots, for example—you can almost be certain that the cat is female.

In Your Notebook *Write three quiz questions about the transmission of human traits and answer them.*

Human Pedigrees

How can pedigrees be used to analyze human inheritance?

Given the complexities of genetics, how would you go about determining whether a trait is caused by a dominant or recessive allele and whether the gene for that trait is autosomal or sex-linked? The answers, not surprisingly, can be found by applying Mendel's basic principles of genetics.

To analyze the pattern of inheritance followed by a particular trait, you can use a chart that shows the relationships within a family. Such a chart is called a **pedigree.** A pedigree shows the presence or absence of a trait according to the relationships between parents, siblings, and offspring. It can be used for any species, not just humans.

The pedigree in **Figure 14–7** shows how one human trait—a white lock of hair just above the forehead—passes through three generations of a family. The allele for the white forelock trait is dominant. At the top of the chart is a grandfather who had the white forelock trait. Two of his three children inherited the trait. Three grandchildren have the trait, but two do not.

BUILD Vocabulary

WORD ORIGINS The word **pedigree** combines the Latin words *pedem,* meaning "foot," and *gruem,* meaning "crane." A crane is a long-legged waterbird. On old manuscripts, a forked sign resembling a crane's footprint indicated a line of ancestral descent.

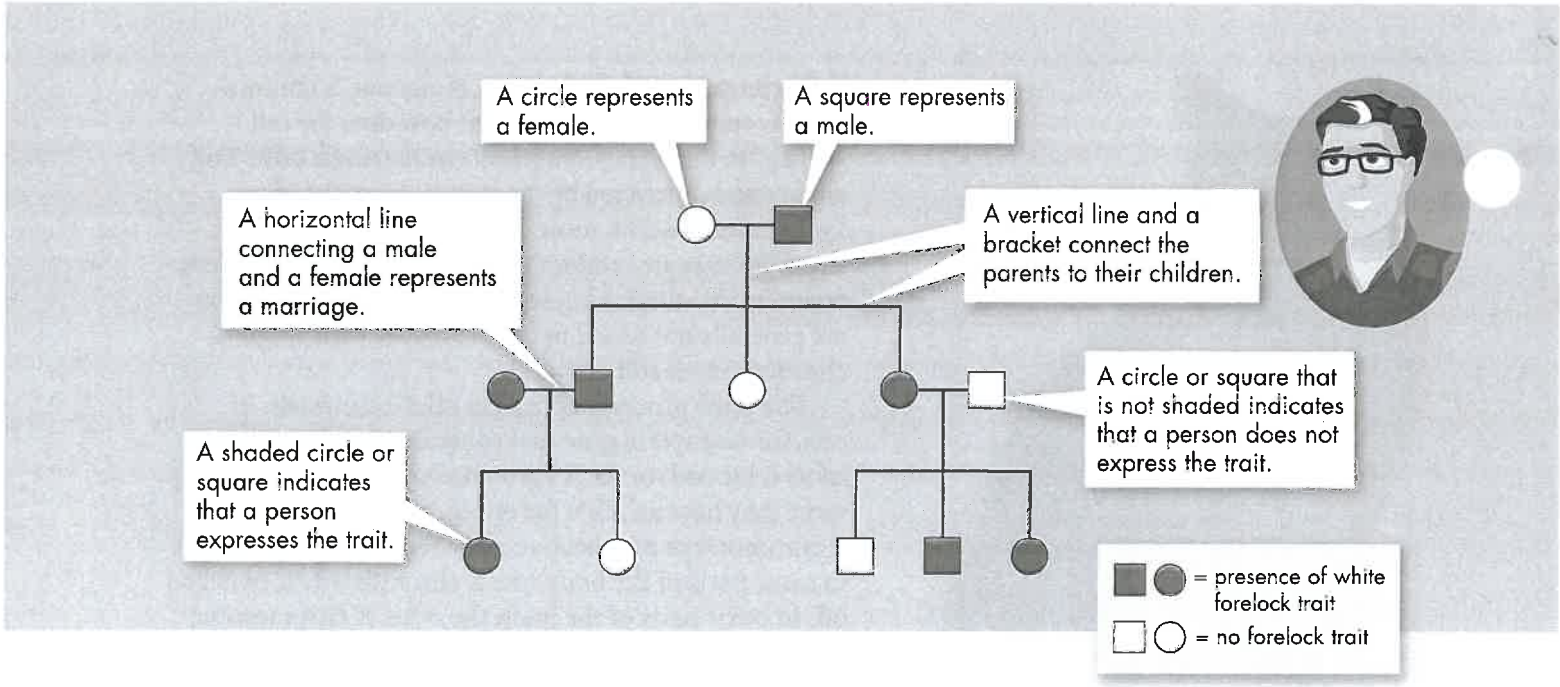

FIGURE 14–7 Pedigree Example
This diagram shows what the symbols in a pedigree represent.
Interpret Visuals ***What are the genotypes of both parents on the left in the second row? How do you know?***

By analyzing a pedigree, we can often infer the genotypes of family members. For example, because the white forelock trait is dominant, all the family members in **Figure 14–7** lacking this trait must have homozygous recessive alleles. One of the grandfather's children lacks the white forelock trait, so the grandfather must be heterozygous for this trait.

With pedigree analysis, it is possible to apply the principles of Mendelian genetics to humans. **The information gained from pedigree analysis makes it possible to determine the nature of genes and alleles associated with inherited human traits.** Based on a pedigree, you can often determine if an allele for a trait is dominant or recessive, autosomal or sex-linked.

14.1 Assessment

Review Key Concepts

1. a. Review What are autosomes?
b. Explain What determines whether a person is male or female?
c. Propose a Solution How can you use karyotypes to identify a species?

2. a. Review Explain how sex-linked traits are inherited.
b. Predict If a woman with type O blood and a man with type AB blood have children, what are the children's possible genotypes?

3. a. Review What does a pedigree show?
b. Infer Why would the Y chromosome be unlikely to contain any of the genes that are absolutely necessary for survival?

VISUAL THINKING

4. Choose a family and a trait, such as facial dimples, that you can trace through three generations. Find out who in the family has had the trait and who has not. Then, draw a pedigree to represent the family history of the trait.

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14.2 Human Genetic Disorders

Key Questions

How do small changes in DNA molecules affect human traits?

What are the effects of errors in meiosis?

Vocabulary

nondisjunction

Taking Notes

Two-Column Chart Before you read, make a two-column chart. In the first column, write three questions you have about genetic disorders. As you read, fill in answers to your questions in the second column. When you have finished, research the answers to your remaining questions.

THINK ABOUT IT Have you ever heard the expression "It runs in the family"? Relatives or friends might have said that about your smile or the shape of your ears, but what could it mean when they talk of diseases and disorders? What, exactly, is a genetic disorder?

From Molecule to Phenotype

How do small changes in DNA molecules affect human traits?

We know that genes are made of DNA and that they interact with the environment to produce an individual organism's characteristics, or phenotype. However, when a gene fails to work or works improperly, serious problems can result.

Molecular research techniques have shown us a direct link between genotype and phenotype. For example, the wax that sometimes builds up in our ear canals can be one of two forms: wet or dry. People of African and European ancestry are more likely to have wet earwax—the dominant form. Those of Asian or Native American ancestry most often have the dry form, which is recessive. A single DNA base in the gene for a membrane-transport protein is the culprit. A simple base change from guanine (G) to adenine (A) causes this protein to produce dry earwax instead of wet earwax.

The connection between molecule and trait, and between genotype and phenotype, is often that simple, and just as direct. **Changes in a gene's DNA sequence can change proteins by altering their amino acid sequences, which may directly affect one's phenotype.** In other words, there is a molecular basis for genetic disorders.

Disorders Caused by Individual Genes

Thousands of genetic disorders are caused by changes in individual genes. These changes often affect specific proteins associated with important cellular functions.

▶ ***Sickle Cell Disease*** This disorder is caused by a defective allele for beta-globin, one of two polypeptides in hemoglobin, the oxygen-carrying protein in red blood cells. The defective polypeptide makes hemoglobin a bit less soluble, causing hemoglobin molecules to stick together when the blood's oxygen level decreases. The molecules clump into long fibers, forcing cells into a distinctive sickle shape, which gives the disorder its name.

Sickle-shaped cells are more rigid than normal red blood cells, and, therefore, they tend to get stuck in the capillaries—the narrowest blood vessels in the body. If the blood stops moving through the capillaries, damage to cells, tissues, and even organs can result.

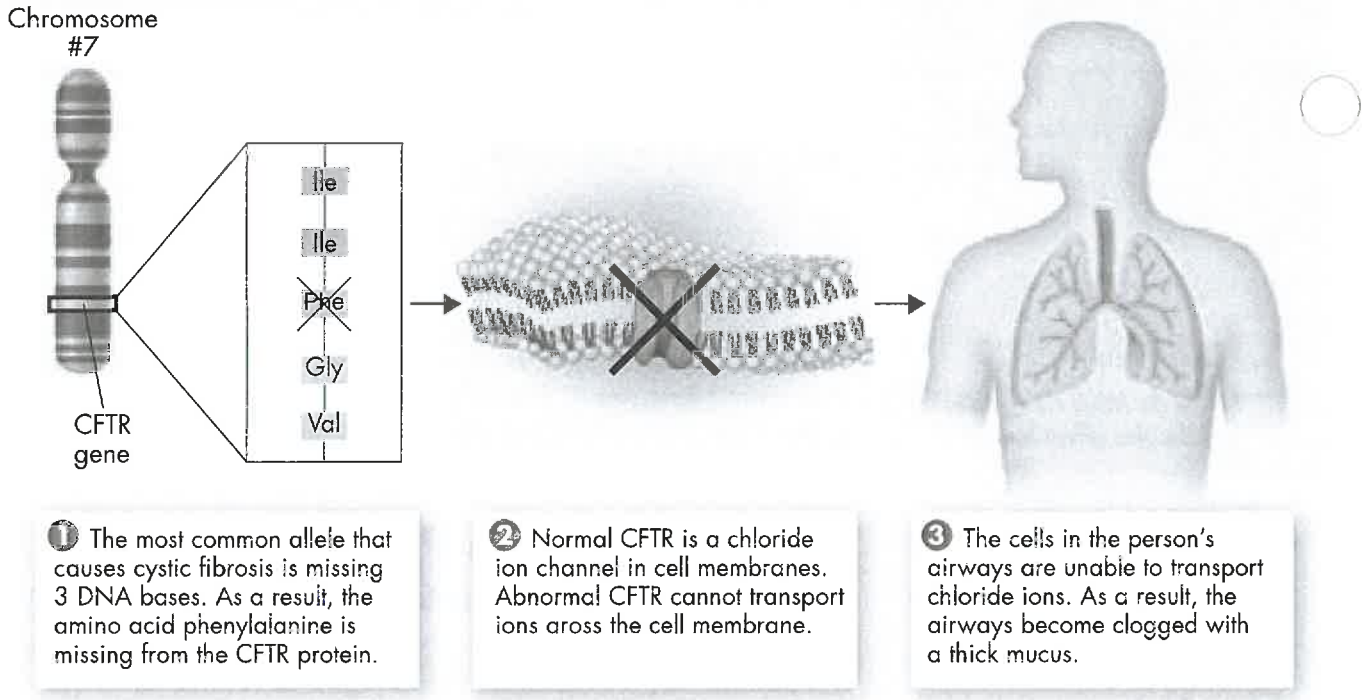


FIGURE 14–8 Mutations Cause Cystic Fibrosis CF is usually caused by the deletion of three bases in the DNA of a single gene. As a result, the body does not produce normal CFTR, a protein needed to transport chloride ions. **Infer** *Why isn't the cause of CF considered a frameshift mutation?*

▶ ***Cystic Fibrosis*** Known as CF for short, cystic fibrosis is most common among people of European ancestry. CF is caused by a genetic change almost as small as the earwax allele. Most cases result from the deletion of just three bases in the gene for a protein called cystic fibrosis transmembrane conductance regulator (CFTR). CFTR normally allows chloride ions (Cl^-) to pass across cell membranes. The loss of these bases removes a single amino acid—phenylalanine—from CFTR, causing the protein to fold improperly. The misfolded protein is then destroyed. With cell membranes unable to transport chloride ions, tissues throughout the body malfunction.

People with one normal copy of the CF allele are unaffected by CF, because they can produce enough CFTR to allow their cells to work properly. Two copies of the defective allele are needed to produce the disorder, which means the CF allele is recessive. Children with CF have serious digestive problems and produce thick, heavy mucus that clogs their lungs and breathing passageways.

▶ ***Huntington's Disease*** Huntington's disease is caused by a dominant allele for a protein found in brain cells. The allele for this disease contains a long string of bases in which the codon CAG—coding for the amino acid glutamine—repeats over and over again, more than 40 times. Despite intensive study, the reason why these long strings of glutamine cause disease is still not clear. The symptoms of Huntington's disease, namely mental deterioration and uncontrollable movements, usually do not appear until middle age. The greater the number of codon repeats, the earlier the disease appears, and the more severe are its symptoms.

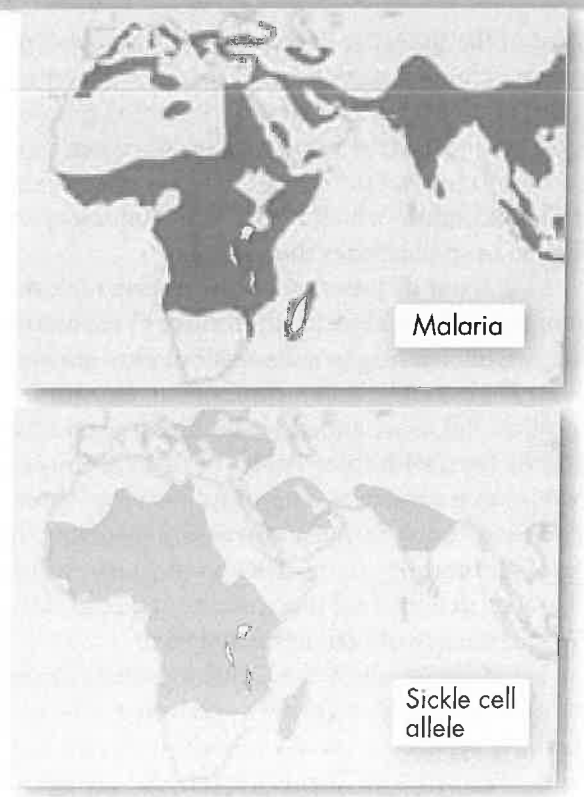
MYSTERY CLUE

Individuals with sickle cell disease have a different amino acid in one of their hemoglobin proteins than people without the disease. What could produce this change?

Analyzing Data

The Geography of Malaria

Malaria is a potentially fatal disease transmitted by mosquitoes. Its cause is a parasite that lives inside red blood cells. The upper map shows the parts of the world where malaria is common. The lower map shows regions where people have the sickle cell allele.



1. Analyze Data What is the relationship between the places where malaria and the sickle cell allele are found?

2. Infer In 1805, a Scottish explorer named Mungo Park led an expedition of European geographers to find the source of the Niger River in Africa. The journey began with a party of 45 Europeans. During the expedition, most of these men perished from malaria. Why do you think their native African guides survived?

3. Form a Hypothesis As the map shows, the sickle cell allele is not found in African populations that are native to southern Africa. Propose an explanation for this discrepancy.

BUILD Vocabulary

WORD ORIGINS The term *malaria* was coined in the mid-eighteenth century from the Italian phrase, *mala aria*, meaning "bad air." It originally referred to the unpleasant odors caused by the release of marsh gases, to which the disease was initially attributed.

Genetic Advantages Disorders such as sickle cell disease and CF are still common in human populations. In the United States, the sickle cell allele is carried by approximately 1 person in 12 of African ancestry, and the CF allele is carried by roughly 1 person in 25 of European ancestry. Why are these alleles still around if they can be fatal for those who carry them? The answers may surprise you.

Most African Americans today are descended from populations that originally lived in west central Africa, where malaria is common. Malaria is a mosquito-borne infection caused by a parasite that lives inside red blood cells. Individuals with just one copy of the sickle cell allele are generally healthy and are also highly resistant to the parasite. This resistance gives them a great advantage against malaria, which even today claims more than a million lives every year.

More than 1000 years ago, the cities of medieval Europe were ravaged by epidemics of typhoid fever. Typhoid is caused by a bacterium that enters the body through cells in the digestive system. The protein produced by the CF allele helps block the entry of this bacterium. Individuals heterozygous for CF would have had an advantage when living in cities with poor sanitation and polluted water, and—because they also carried a normal allele—these individuals would not have suffered from cystic fibrosis.

Chromosomal Disorders

What are the effects of errors in meiosis?

Most of the time, the process of meiosis works perfectly and each human gamete gets exactly 23 chromosomes. Every now and then, however, something goes wrong. The most common error in meiosis occurs when homologous chromosomes fail to separate. This mistake is known as **nondisjunction,** which means "not coming apart." **Figure 14–9** illustrates the process.

If nondisjunction occurs during meiosis, gametes with an abnormal number of chromosomes may result, leading to a disorder of chromosome numbers. For example, if two copies of an autosomal chromosome fail to separate during meiosis, an individual may be born with three copies of that chromosome. This condition is known as a trisomy, meaning "three bodies." The most common form of trisomy, involving three copies of chromosome 21, is Down syndrome, which is often characterized by mild to severe mental retardation and a high frequency of certain birth defects.

Nondisjunction of the X chromosomes can lead to a disorder known as Turner's syndrome. A female with Turner's syndrome usually inherits only one X chromosome. Women with Turner's syndrome are sterile, which means that they are unable to reproduce. Their sex organs do not develop properly at puberty.

In males, nondisjunction may cause Klinefelter's syndrome, resulting from the inheritance of an extra X chromosome, which interferes with meiosis and usually prevents these individuals from reproducing. There have been no reported instances of babies being born without an X chromosome, indicating that this chromosome contains genes that are vital for the survival and development of the embryo.

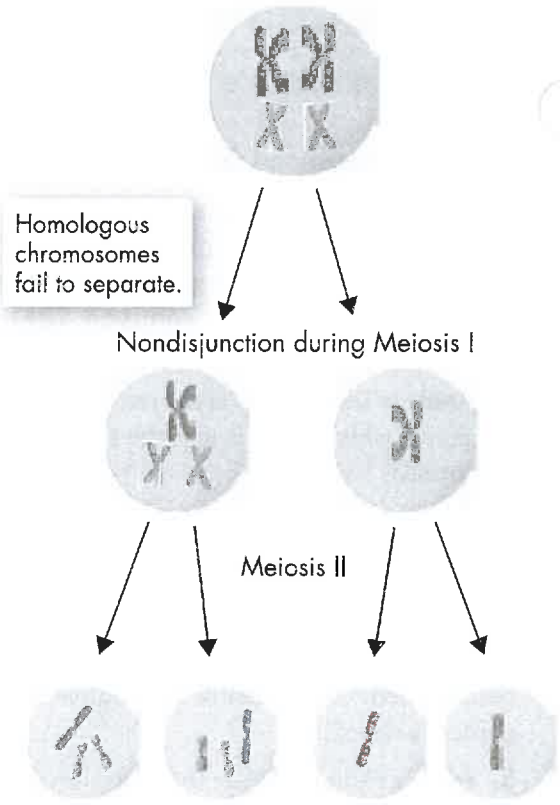


FIGURE 14–9 Nondisjunction This failure of meiosis causes gametes to have an abnormal number of chromosomes. **Apply Concepts** ***Which phase of meiosis is shown in the first cell?***

14.2 Assessment

Review Key Concepts

1. a. Review How can a small change in a person's DNA cause a genetic disorder?

b. Infer How do genetic disorders such as CF support the theory of evolution?

2. a. Review Describe two sex chromosome disorders.

b. Apply Concepts How does nondisjunction cause chromosomal disorders?

WRITE ABOUT SCIENCE

Description

3. Write a paragraph explaining the process of nondisjunction. (*Hint:* To organize your writing, create a flowchart that shows the steps in the process.)

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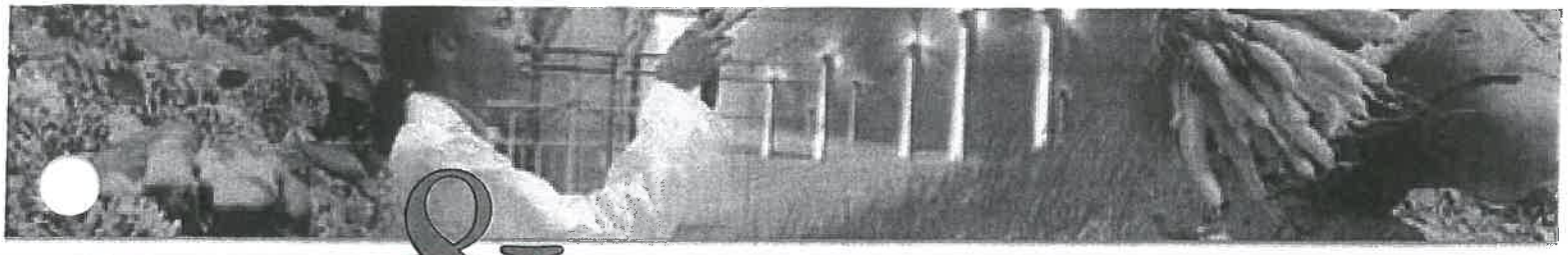
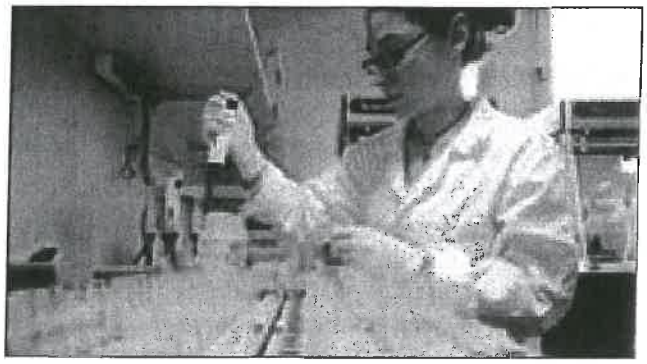


Biology & Society

Are Laws Protecting Genetic Privacy Necessary?

The rapid development of new tools and techniques to analyze DNA makes it possible to test for alleles related to thousands of medical conditions. In theory, the results of genetic testing should benefit everyone. Accurate genetic data helps physicians select the proper treatments for patients. It may allow people with genes that place them at risk of certain conditions to minimize those risks.

At issue, however, is individual privacy. Once a test is done, who has access to the data, and how can they use it? Could employers refuse to hire people who might drive up their medical costs? Might insurance companies refuse to renew the policies of individuals with genes for certain disorders? These are not hypothetical questions. In 2005, managers of a professional basketball team asked one of its players to be tested for a gene linked to heart ailments. When he refused, they traded the player to another team. Dr. Francis Collins, director of the National Human Genome Research Institute, worries that "the public is afraid of taking advantage of genetic testing." Is he correct? Should genetic data be protected by law, or should it be open to public view?



Many commercial laboratories test human DNA for genetic disorders.

The Viewpoints

Genetic Privacy Does Not Need Legal Protection

Other laws already protect individuals from discrimination on the basis of medical disability. Employers and insurance companies are nonetheless allowed to ask individuals if they smoke, use alcohol, or have a history of medical problems. Having this information allows employers to make intelligent choices about whom to hire. It also helps insurance companies maintain lower rates for their healthiest clients. Free access to genetic data should be a public right.

Genetic Privacy Should Be Protected by Law

The Genetic Information Nondiscrimination Act (GINA) went into effect in 2009, and it provides important protections to personal privacy. Individuals may not take advantage of today's advances in genetic medicine if they fear their personal information might be used to deny them employment or insurance. We need such laws to realize the full benefits of modern medicine and to protect otherwise healthy individuals from genetic discrimination.

Research and Decide

1. Analyze the Viewpoints To make an informed decision, learn more about genetic testing by consulting library or Internet resources. Then, list the key arguments expressed by the proponents and critics of both points of view. Find out if laws preventing genetic discrimination have been proposed or passed in your state.

2. Form an Opinion Should access and use of genetic data be regulated? Weigh both sides of the issue. Who will benefit from the sharing of genetic data? Will anyone suffer? Do some arguments outweigh others? If so, which ones? Explain your answers.

Chapter 2

Basic Cell Biology

Essential Grade 9 Review!

In This Chapter

- Getting to know the cell
- Understanding chromosomes
- Exploring simple cell division
- Appreciating the complexities of meiosis

Genetics and the study of how cells work are closely related. The process of passing genetic material from one generation to the next depends completely on how cells grow and divide. To reproduce, a simple organism such as bacteria or yeast simply copies its DNA (through a process called *replication,* which I cover in Chapter 7) and splits in two. But organisms that reproduce sexually go through a complicated dance that includes mixing and matching strands of DNA (a process called *recombination*) and then halving the amount of DNA for special sex cells, allowing completely new genetic combinations for their offspring. These amazing processes are part of what makes you unique. So come inside your cell — you need to be familiar with the processes of *mitosis* (cell division) and *meiosis* (the production of sex cells) to appreciate how genetics works.

Looking Around Your Cell

There are two basic kinds of organisms:

- **Prokaryotes:** Organisms whose cells lack a nucleus and therefore have DNA floating loosely in the liquid center of the cell
- **Eukaryotes:** Organisms that have a well-defined nucleus to house and protect the DNA

A *nucleus* is a compartment filled with DNA surrounded by a membrane.

The basic biologies of prokaryotes and eukaryotes are similar but not identical. Because all living things fall into these two groups, understanding the differences and similarities between cell types is important. In this section, I

show you how to distinguish the two kinds of cells from each other, and you get a quick tour of the insides of cells — both with and without nuclei. Figure 2-1 shows you the structure of each type of cell.

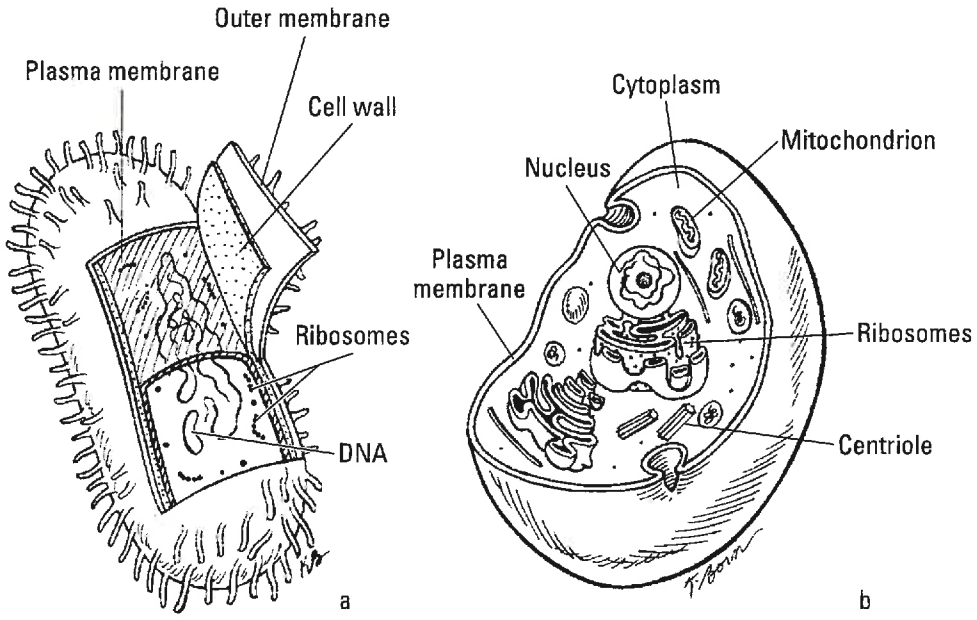


Figure 2-1: A prokaryotic cell (a) is very simple compared to a eukaryotic cell (b).

Cells without a nucleus

Scientists classify organisms composed of cells without nuclei as *prokaryotes,* which means "before nucleus." Prokaryotes are the most common forms of life on earth. You are, at this very moment, covered in and inhabited by millions of prokaryotic cells: bacteria. Much of your life and your body's processes depend on these arrangements; for example, the digestion going on in your intestines is partially powered by bacteria that break down the food you eat. Most of the bacteria in your body are completely harmless, but some species of bacteria can be vicious and deadly, causing rapidly transmitted diseases such as cholera.

All bacteria, regardless of temperament, are simple, one-celled, prokaryotic organisms. None has cell nuclei, and all are small cells with relatively small amounts of DNA (see Chapter 8 for more on the amounts of DNA different organisms possess).

The exterior of a prokaryotic cell is encapsulated by a *cell wall* that serves as the bacteria's only protection from the outside world. A *plasma membrane* (*membranes* are thin sheets or layers) regulates the exchange of nutrients, water, and gases that nourish the bacterial cell. DNA, usually in the form of a single, hoop-shaped piece, floats around inside the cell; segments of DNA

like this one are called *chromosomes* (see the section "Examining the basics of chromosomes" later in the chapter). The liquid interior of the cell is called the *cytoplasm.* The cytoplasm provides a cushiony, watery home for the DNA and other cell machinery that carry out the business of living. Prokaryotes divide, and thus reproduce, by simple mitosis, which I cover in detail in the "Mitosis: Splitting Up" section later in the chapter.

Cells with a nucleus

Scientists classify organisms that have cells with nuclei as *eukaryotes,* which means "true nucleus." Eukaryotes range in complexity from simple, one-celled animals and plants to complex, multicellular organisms like you. Eukaryotic cells are fairly complicated and have numerous parts to keep track of (refer to Figure 2-1). Like prokaryotes, eukaryotic cells are held together by a *plasma membrane,* and sometimes a *cell wall* surrounds the membrane (plants, for example, have cell walls). But that's where the similarities end.

The most important feature of the eukaryotic cell is the *nucleus* — the membrane-surrounded compartment that houses the DNA that's divided into one or more chromosomes. The nucleus protects the DNA from damage during day-to-day living. Eukaryotic chromosomes are usually long, string-like segments of DNA instead of the hoop-shaped ones found in prokaryotes. Another hallmark of eukaryotes is the way the DNA is packaged: Eukaryotes usually have much larger amounts of DNA than prokaryotes, and to fit all that DNA into the tiny cell nucleus, it must be tightly wound around special proteins. (For all the details about DNA packaging for eukaryotes, flip to Chapter 6.)

Unlike prokaryotes, eukaryotes have all sorts of cell parts, called *organelles,* that help carry out the business of living. The organelles float around in the watery cytoplasm outside the nucleus. Two of the most important organelles are

- **Mitochondria:** The powerhouses of the eukaryotic cell, mitochondria pump out energy by converting glucose to ATP (adenosine triphosphate). ATP acts like a battery of sorts, storing energy until it's needed for day-to-day living. Both animals and plants have mitochondria.
- **Chloroplasts:** These organelles are unique to plants. They process the energy of sunlight into sugars that the plant mitochondria use to generate the energy that nourishes the living cells.

Eukaryotic cells are able to carry out behaviors that prokaryotes can't. For example, one-celled eukaryotes often have appendages, such as long tails (called *flagella*) or hair-like projections (called *cilia*), that work like hundreds of tiny paddles, helping them move around. Also, only eukaryotic cells are capable of ingesting fluids and particles for nutrition; prokaryotes must transport materials through their cell walls, a process that severely limits their dietary options.

In most multicellular eukaryotes, cells come in two basic varieties: body cells (called *somatic* cells) or sex cells. The two cell types have different functions and are produced in different ways.

Somatic cells

Somatic cells are produced by simple cell division called *mitosis* (see the section "Mitosis: Splitting Up" for details). Somatic cells of multicellular organisms like humans are differentiated into special cell types. Skin cells and muscle cells are both somatic cells, for instance, but if you were to examine your skin cells under a microscope and compare them with your muscle cells, you'd see that their structures are very different. The various cells that make up your body all have the same basic components (membrane, organelles, and so on), but the arrangements of the elements change from one cell type to the next so that they can carry out various jobs such as digestion (intestinal cells), energy storage (fat cells), or oxygen transport to your tissues (blood cells).

Sex cells

Sex cells are specialized cells used for reproduction. Only eukaryotic organisms engage in sexual reproduction, which I cover in detail at the end of this chapter in the section "Mommy, where did I come from?" *Sexual reproduction* combines genetic material from two organisms and requires special preparation in the form of a reduction in the amount of genetic material allocated to sex cells — a process called *meiosis* (see "Meiosis: Making Cells for Reproduction" later in the chapter for an explanation). In humans, the two types of sex cells are eggs and sperm.

Examining the basics of chromosomes

Chromosomes are threadlike strands composed of DNA. To pass genetic traits from one generation to the next, the chromosomes must be copied (see Chapter 7), and then the copies must be divvied up. Most prokaryotes have only one circular chromosome that, when copied, is passed on to the *daughter cells* (new cells created by cell division) during mitosis. Eukaryotes have more complex problems to solve (like divvying up half of the chromosomes to make sex cells), and their chromosomes behave differently during mitosis and meiosis. Additionally, various scientific terms describe the anatomy, shapes, number of copies, and situations of eukaryotic chromosomes. This section gets into the intricacies of chromosomes in eukaryotic cells because they're so complex.

Counting out chromosome numbers

Each eukaryotic organism has a specific number of chromosomes per cell — ranging from one to many. For example, humans have 46 total chromosomes. These chromosomes come in two varieties:

- **Sex chromosomes:** These chromosomes determine gender. Human cells contain two sex chromosomes. If you're female, you have two X chromosomes, and if you're male, you have an X and a Y chromosome. (To find out more about how sex is determined by the X and Y chromosomes, flip to Chapter 5.)
- **Autosomal chromosomes:** *Autosomal* simply refers to non-sex chromosomes. Sticking with the human example, if you do the math, you can see that humans have 44 autosomal chromosomes.

Ah, but there's more. In humans, chromosomes come in pairs. That means you have 22 pairs of uniquely shaped autosomal chromosomes plus 1 pair of sex chromosomes for a total of 23 chromosome pairs. Your autosomal chromosomes are identified by numbers — 1 through 22. So you have two chromosome 1s, two 2s, and so on. Figure 2-2 shows you how all human chromosomes are divided into pairs and numbered. (A *karyotype* like the one pictured in Figure 2-2 is one way chromosomes are examined; you discover more about karyotyping in Chapter 15.)

When chromosomes are sorted into pairs, the individual chromosomes in each pair are considered *homologous,* meaning that the paired chromosomes are identical to one another according to which genes they carry. In addition, your homologous chromosomes are identical in shape and size. These pairs of chromosomes are sometimes referred to as *homologs* for short.

Chromosome numbers can be a bit confusing. Humans are *diploid,* meaning we have two copies of each chromosome. Some organisms (like bees and wasps) have only one set of chromosomes (cells with one set of chromosomes are called *haploid*); others have three, four, or as many as sixteen copies of each chromosome! The number of chromosome sets held by a particular organism is called the *ploidy.* For more on chromosome numbers, see Chapter 15.

The total number of chromosomes doesn't tell you what the ploidy of an organism is. For that reason, the number of chromosomes an organism has is often listed as some multiple of *n.* A single set of chromosomes referred to by the *n* is the haploid number. Humans are $2n = 46$ (indicating that humans are diploid and their total number of chromosomes is 46). Human sex cells such as eggs or sperm are haploid (see "Mommy, where did I come from?" later in this chapter).

Geneticists believe that the homologous pairs of chromosomes in humans started as one set (that is, *haploid*), and the entire set was duplicated at some point in some distant ancestor, many millions of years ago.

Examining chromosome anatomy

Chromosomes are often depicted in stick-like forms, like those you see in Figure 2-3. Chromosomes don't look like sticks, though. In fact, most of the time they're loose and string-like. Chromosomes only take on this distinctive

shape and form when cell division is about to take place (during metaphase of meiosis or mitosis). They're often drawn this way so that the special characteristics of eukaryotic chromosomes are easier to see. Figure 2-3 points out the important features of eukaryotic chromosomes.

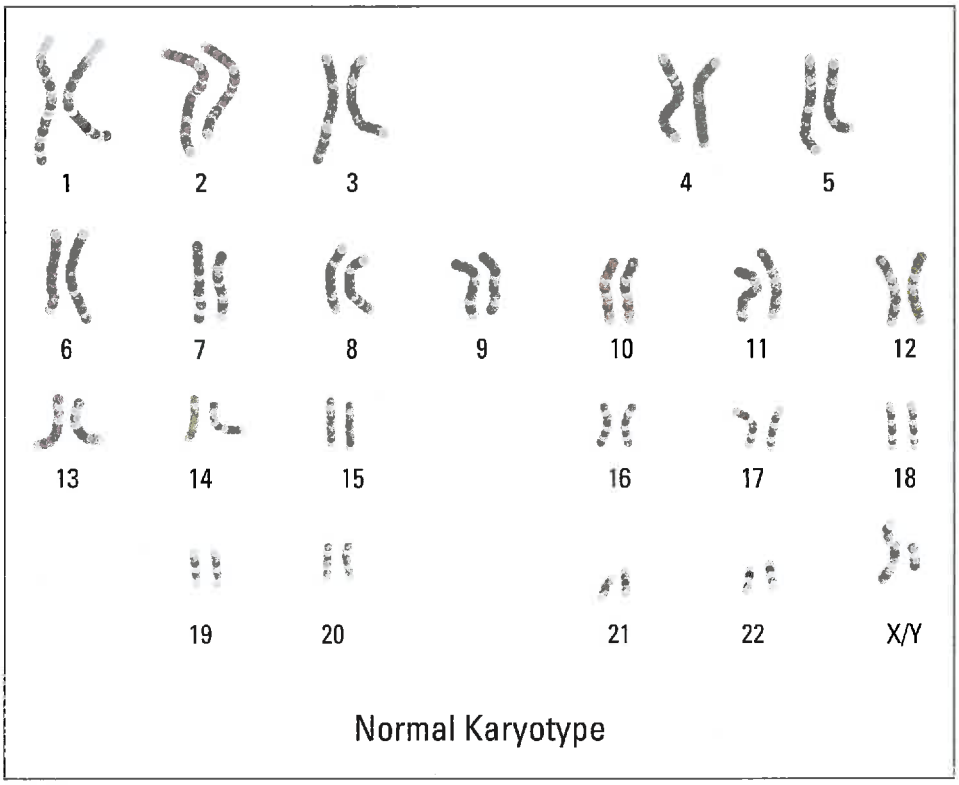


Figure 2-2: The 46 human chromosomes are divided into 23 pairs.

The part of the chromosome that appears pinched (in Figure 2-3, located in the middle of the chromosomes) is called the *centromere*. The placement of the centromere (whether it's closer to the top, middle, or bottom of the chromosome; see Figure 2-4) is what gives each chromosome its unique shape. The ends of the chromosomes are called *telomeres*. Telomeres are made of densely packed DNA and serve to protect the DNA message that the chromosome carries. (Flip to Chapter 23 for more about telomeres and how they may affect the process of aging.)

The differences in shapes and sizes of chromosomes are easy to see, but the most important differences between chromosomes are hidden deep inside the DNA. Chromosomes carry *genes* — sections of DNA that make up the building plans for physical traits. The genes tell the body how, when, and where to make all the structures that are necessary for the processes of living (for more on how genes work, flip to Chapter 11). Each pair of homologous chromosomes carries the same — but not necessarily identical — genes. For example,

both chromosomes of a particular homologous pair may contain the gene for hair color, but one can be a "brown hair" version of the gene — alternative versions of genes are called *alleles* (refer to Figure 2-3) — and the other can be a "blond hair" allele.

Any given gene can have one or more alleles. In Figure 2-3, one chromosome carries the allele *A* while its homolog carries the allele *a* (the relative size of an allele is very small; the alleles are large here so you can see them). The alleles code for the different physical traits *(phenotypes)* you see in animals and plants, like hair color or flower shape. You can find out more about how alleles affect phenotype in Chapter 3.

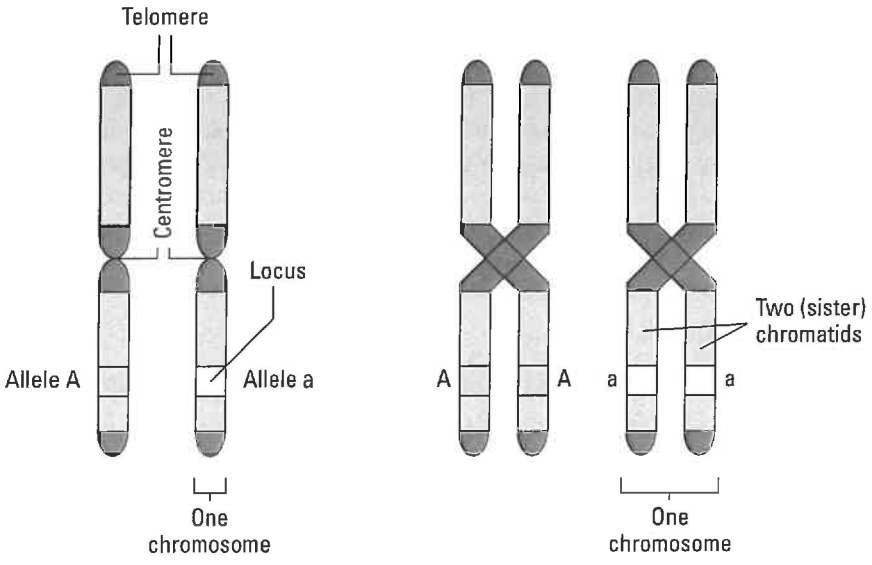


Figure 2-3: Basic structure of eukaryotic chromosomes.

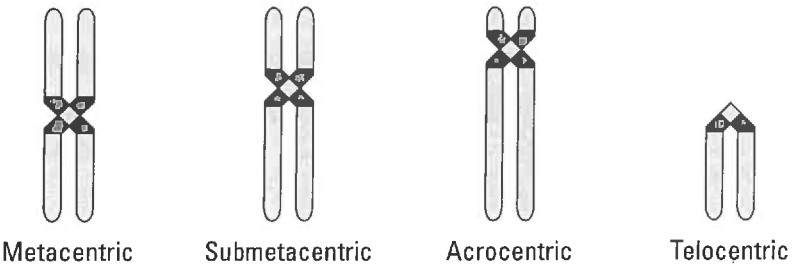


Figure 2-4: Chromosomes are classified based on the locations of their centromeres.

Each point along the chromosome is called a *locus* (Latin for "place"). The plural of locus is *loci* (pronounced *low*-sigh). Most of the phenotypes that you see are produced by multiple genes (that is, genes occurring at different loci and often on different chromosomes) acting together. For instance, human

eye color is determined by at least three genes that reside on two different chromosomes. You can find out more about how genes are arranged along chromosomes in Chapter 15.

Mitosis: Splitting Up

Most cells have simple lifestyles: They grow, divide, and eventually die. Figure 2-5 illustrates the basic life cycle of a typical somatic, or body, cell.

The *cell cycle* (the stages a cell goes through from one division to another) is tightly regulated; some cells divide all the time, and others never divide at all. Your body uses mitosis to provide new cells when you grow and to replace cells that wear out or become damaged from injury. Talk about multitasking — you're going through mitosis right now, while you read this book! Some cells divide only part of the time, when new cells are needed to handle certain jobs like fighting infection. Cancer cells, on the other hand, get carried away and divide too often. (In Chapter 14, you can find out how the cell cycle is regulated and what happens when it goes awry.)

The cell cycle includes *mitosis* — the process of reproducing the cell nucleus by division. The result of each round of the cell cycle is a simple cell division that creates two identical new cells from one original cell. During mitosis, all DNA present in the cell is copied (see Chapter 7), and when the original cell divides, a complete collection of all the chromosomes (in humans, 23 pairs) goes to each of the two resulting cells. Prokaryotes and some simple eukaryotic organisms use mitosis to reproduce themselves. (More complex eukaryotic organisms use *meiosis* for sexual reproduction, in which each of the two sex cells sends only one copy of each chromosome into the eggs or sperm. You can read all about that in the section "Meiosis: Making Cells for Reproduction" later in this chapter.)

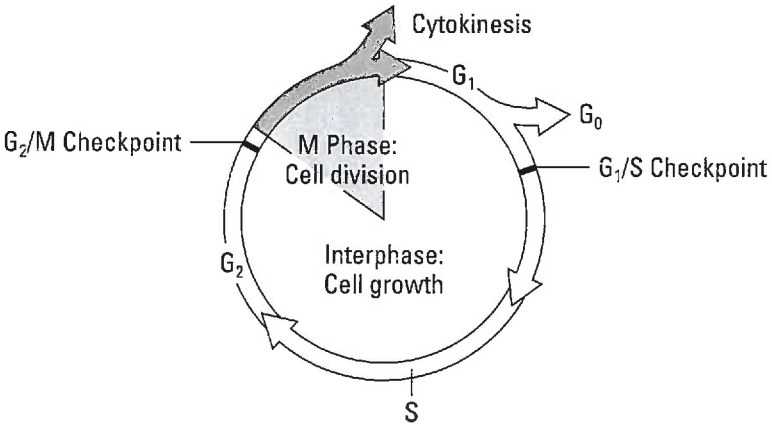


Figure 2-5: The cell cycle: mitosis, cell division, and all points in between.

You should remember two important points about mitosis:

- **Mitosis produces two identical cells.** The new cells are identical to each other *and* to the cell that divided to create them.
- **Cells created by mitosis have exactly the same number of chromosomes as the original cell did.** If the original cell had 46 chromosomes, the new cells each have 46 chromosomes.

Mitosis is only one of the major phases in the cell cycle; the other is *interphase.* In the following sections, I guide you through the phases of the cell cycle and tell you exactly what happens during each one.

Step 1: Time to grow

Interphase is the part of the cell cycle during which the cell grows, copies its DNA, and prepares to divide. Interphase occurs in three stages: the G1 phase, the S phase, and the G2 phase.

G1 phase

When a cell begins life, such as the moment an egg is fertilized, the first thing that happens is the original cell starts to grow. This period of growth is called the *G1 phase* of interphase. Lots of things happen during G1: DNA supervises the work of the cell, *metabolism* (the exchange of oxygen and carbon dioxide) occurs, and cells breathe and "eat."

Some cells opt out of the cell cycle permanently, stop growing, and exit the process at G_0. Your brain cells, for example, have retired from the cell cycle. Mature red blood cells and muscle cells don't divide, either. In fact, human red blood cells have no nuclei and thus possess no DNA of their own.

If the cell in question plans to divide, though, it can't stay in G1 forever. Actively dividing cells go through the whole cell cycle every 24 hours or so. After a predetermined period of growth that lasts from a few minutes to several hours, the cell arrives at the first checkpoint (refer to Figure 2-5). When the cell passes the first checkpoint, there's no turning back.

Various proteins control when the cell moves from one phase of the cycle to the next. At the first checkpoint, proteins called *cyclins* and enzymes called *kinases* control the border between G1 and the next phase. Cyclins and kinases interact to cue up the various stages of the merry-go-round of cell division. Two particular chemicals, CDK (cyclin dependent kinase) and G1 cyclin, hook up to escort the cell over the border from G1 to S — the next phase.

S phase

S phase is the point at which the cell's DNA is replicated (here, *S* refers to *synthesis,* or copying, of the DNA). When the cell enters the S phase, activity around the chromosomes really steps up. All the chromosomes must be copied to make exact replicas that later are passed on to the newly formed daughter cells produced by cell division. DNA replication is a very complex process that gets full coverage in Chapter 7.

For now, all you need to know is that all the cell's chromosomes are copied during S, and the copies stay together as a unit (joined at the centromere; refer back to Figure 2-3) when the cell moves from S into G2 — the final step in interphase. The replicated chromosomes are called *sister chromatids* (refer to Figure 2-3), which are alike in every way. They carry the exact same copies of the exact same genes. During mitosis (or meiosis), the sister chromatids are divided up and sent to the daughter cells as part of the cell cycle.

G2 phase

The *G2 phase* leads up to cell division. It's the last phase before actual mitosis gets underway. G2, sometimes called *Gap 2,* gives the cell time to get bigger before splitting into two smaller cells. Another set of cyclins and CDK work together to push the cell through the second checkpoint located at the border between G2 and mitosis. (For details on the first checkpoint, jump back to the section "G1 phase.") As the cell grows, the chromosomes, now copied and hooked together as sister chromatids, stay together inside the cell nucleus. (The DNA is still "relaxed" at this point and hasn't yet taken on the fat, sausage-shaped appearance it assumes during mitosis.) After the cell crosses the G2/M checkpoint (refer to Figure 2-5), the business of mitosis formally gets underway.

Step 2: Divvying up the chromosomes

In the cell cycle, *mitosis* is the process of dividing up the newly copied chromosomes (that were created in interphase; see the preceding section) to make certain that the new cells each get a full set. Generally, mitosis is divided into four phases, which you can see in Figure 2-6 and read about in the following sections.

The phases of mitosis are a bit artificial, because the movement doesn't stop at each point; instead, the chromosomes cruise right from one phase to the next. But dividing the process into phases is useful for understanding how the chromosomes go from being all mixed together to neatly parting ways and getting into the proper, newly formed cells.

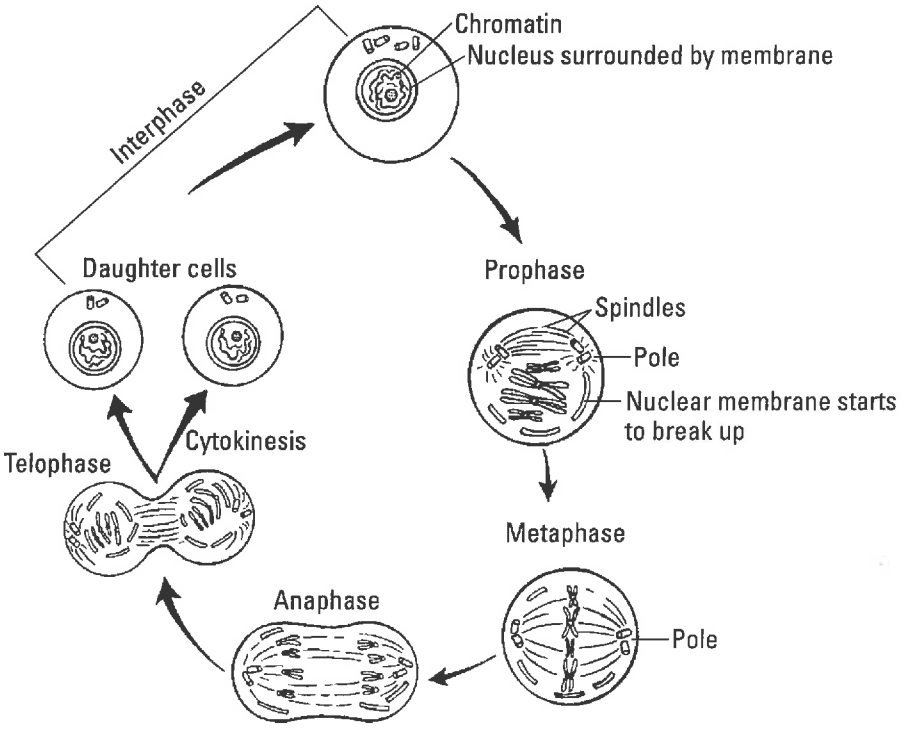


Figure 2-6: The process of mitosis, broken down into four stages: prophase, metaphase, anaphase, and telophase.

Prophase

During *prophase,* the chromosomes get very compact and condensed, taking on the familiar sausage shape. During interphase (see the "Step 1: Time to grow" section earlier in this chapter), the DNA that makes up the chromosomes is tightly wound around special proteins, sort of like string wrapped around beads. The whole "necklace" is wound tightly on itself to compress the enormous DNA molecules to sizes small enough to fit inside the cell nucleus. But even when coiled during interphase, the chromosomes are still so threadlike and tiny that they're essentially invisible. That changes during prophase, when the chromosomes become so densely packed that you can easily see them with an ordinary light microscope.

By the time they reach prophase, chromosomes have duplicated to form sister chromatids (refer to Figure 2-3). Sister chromatids of each chromosome are exact twin copies of each other. Each chromatid is actually a chromosome in its own right, but thinking of chromosomes as chromatids may help you keep all the players straight during the process of division.

As the chromosomes/chromatids condense, the cell nucleus starts breaking up, allowing the chromosomes to move freely across the cell as the process of cell division progresses.

Metaphase

Metaphase is the point when the chromosomes all line up in the center of the cell. After the nuclear membrane dissolves and prophase is complete, the chromosomes go from being a tangled mass to lining up in a more or less neat row in the center of the cell (refer to Figure 2-6). Threadlike strands called *spindles* grab each chromosome around its waist-like centromere. The spindles are attached to points on either side of the cell called *poles*.

Sometimes, scientists use geographic terms to describe the positions of chromosomes during metaphase: The chromosomes line up at the equator and are attached to the poles. This trick may help you better visualize the events of metaphase.

Anaphase

During *anaphase,* the sister chromatids are pulled apart, and the resulting halves migrate to opposite poles (refer to Figure 2-6). At this point, it's easy to see that the chromatids are actually chromosomes. Every sister chromatid gets split apart so that the cell that's about to be formed ends up with a full set of all the original cell's chromosomes.

Telophase

Finally, during *telophase,* nuclear membranes begin to form around the two sets of separated chromosomes (refer to Figure 2-6). The chromosomes begin to relax and take on their usual interphase form. The cell itself begins to divide as telophase comes to an end.

Step 3: The big divide

When mitosis is complete and new nuclei have formed, the cell divides into two smaller, identical cells. The division of one cell into two is called *cytokinesis* (*cyto* meaning "cell" and *kinesis* meaning "movement"). Technically, cytokinesis happens after metaphase is over and before interphase begins. Each new cell has a full set of chromosomes, just as the original cell did. All the organelles and cytoplasm present in the original cell are divided up to provide the new cell with all the machinery it needs for metabolism and growth. The new cells are now at interphase (specifically, the G1 stage) and are ready to begin the cell cycle again.

Meiosis: Making Cells for Reproduction

Meiosis is a cell division that includes reducing the chromosome number as preparation for sexual reproduction. Meiosis reduces the amount of DNA by half so that when fertilization occurs, each offspring gets a full set

of chromosomes. As a result of meiosis, the cell goes from being diploid to being haploid. Or, to put it another way, the cell goes from being *2n* to being *n*. In humans, this means that the cells produced by meiosis (either eggs or sperm) have 23 chromosomes each — one copy of each of the homologous chromosomes. (See the section "Counting out chromosome numbers" earlier in this chapter for more information.)

Meiosis has many characteristics in common with mitosis. The stages go by similar names, and the chromosomes move around similarly, but the products of meiosis are completely different from those of mitosis. Whereas mitosis ends with two identical cells, meiosis produces *four* cells each with *half* the amount of DNA that the original cell contained. Furthermore, with meiosis, the homologous chromosomes go through a complex exchange of segments of DNA called *recombination*. Recombination is one of the most important aspects of meiosis and leads to genetic variation that allows each individual produced by sexual reproduction to be truly unique.

Meiosis goes through two rounds of division: meiosis I and the sequel, meiosis II. Figure 2-7 shows the progressing stages of both meiosis I and meiosis II. Unlike lots of movie sequels, the sequel in meiosis is really necessary. In both rounds of division, the chromosomes go through stages that resemble those in mitosis. However, the chromosomes undergo different actions in meiotic prophase, metaphase, anaphase, and telophase.

Students often get stuck on the phases of meiosis and miss its most important aspects: recombination and the division of the chromosomes. To prevent that sort of confusion, I don't break down meiosis by phases. Instead, I focus on the activities of the chromosomes themselves.

In meiosis I:

- The homologous pairs of chromosomes line up side by side and exchange parts. This is called *crossing-over* or *recombination,* and it occurs during prophase I.
- During metaphase I, the homologous chromosomes line up at the equator of the cell (called the *metaphase plate*), and homologs go to opposite poles during the first round of anaphase.
- The cell divides in telophase I, reducing the amount of genetic material by half, and enters a second round of division — meiosis II.

During meiosis II:

- The individual chromosomes (as sister chromatids) condense during prophase II and line up at the metaphase plates of both cells (metaphase II).
- The chromatids separate and go to opposite poles (anaphase II).
- The cells divide, resulting in a total of *four* daughter cells, each possessing *one* copy of each chromosome.

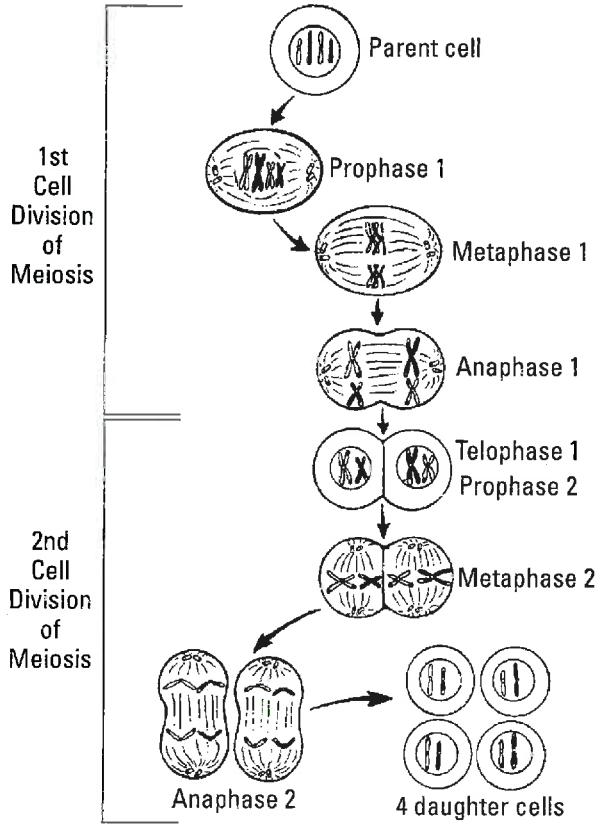


Figure 2-7: The phases of meiosis.

Meiosis part 1

Cells that undergo meiosis start in a phase similar to the interphase that precedes mitosis. The cells grow in a G1 phase, undergo DNA replication during S, and prepare for division during G2. (To review what happens in each of these phases, flip back to the section "Step 1: Time to grow.") When meiosis is about to begin, the chromosomes condense. By the time meiotic interphase is complete, the chromosomes have been copied and are hitched up as sister chromatids, just as they would be in mitosis. Next up are the phases of meiosis I, which I profile in the sections that follow.

Find your partner

During prophase I (labeled "I" because it's in the first round of meiosis), the homologous chromosomes find each other. These homologous chromosomes originally came from the mother and father of the individual whose cells are now undergoing meiosis. Thus, during meiosis, maternal and paternal chromosomes, as homologs, line up side by side. In Figure 2-2, you can see an entire set of 46 human chromosomes. Although the members of

the pair seem identical, they're not. The homologous chromosomes have different combinations of alleles at the thousands of loci along each chromosome. (For more on alleles, jump to the section "Examining chromosome anatomy" earlier in this chapter.)

Recombining makes you unique

When the homologous chromosomes pair up in prophase I, the chromatids of the two homologs actually zip together, and the chromatids exchange parts of their arms. Enzymes cut the chromosomes into pieces and seal the newly combined strands back together in an action called *crossing-over*. When crossing-over is complete, the chromatids consist of part of their original DNA and part of their homolog's DNA. The loci don't get mixed up or turned around — the chromosome sequence stays in its original order. The only thing that's different is that the maternal and paternal chromosomes (as homologs) are now mixed together.

Figure 2-8 illustrates crossing-over in action. The figure shows one pair of homologous chromosomes and two loci. At both loci, the chromosomes have alternative forms of the genes. In other words, the alleles are different: Homolog one has *A* and *b*, and homolog two has *a* and *B*. When replication takes place, the sister chromatids are identical (because they're exact copies of each other). After crossing-over, the two sister chromatids have exchanged arms. Thus, each homolog has a sister chromatid that's different.

Partners divide

The recombined homologs line up at the metaphase equator of the cell (refer to Figure 2-7). The nuclear membrane begins to break down, and in a process similar to mitotic anaphase, spindle fibers grasp the homologous chromosomes by their centromeres and pull them to opposite sides of the cell.

At the end of the first phase of meiosis, the cell undergoes its first round of division (telophase 1, followed by cytokinesis 1). The newly divided cells each contain one set of chromosomes, the now partnerless homologs, still in the form of replicated sister chromatids.

When the homologs line up, maternal and paternal chromosomes pair up, but it's a tossup as to which side of the equator each one ends up on. Therefore, each pair of homologs divides independently of every other homologous pair. This is the basis of the principle of independent assortment, which I cover in Chapters 3 and 4.

Following telophase I, the cells enter an in-between round called *interkinesis* (which means "between movements"). The chromosomes relax and lose their fat, ready-for-metaphase appearance. Interkinesis is just a "resting" phase in preparation for the second round of meiosis.

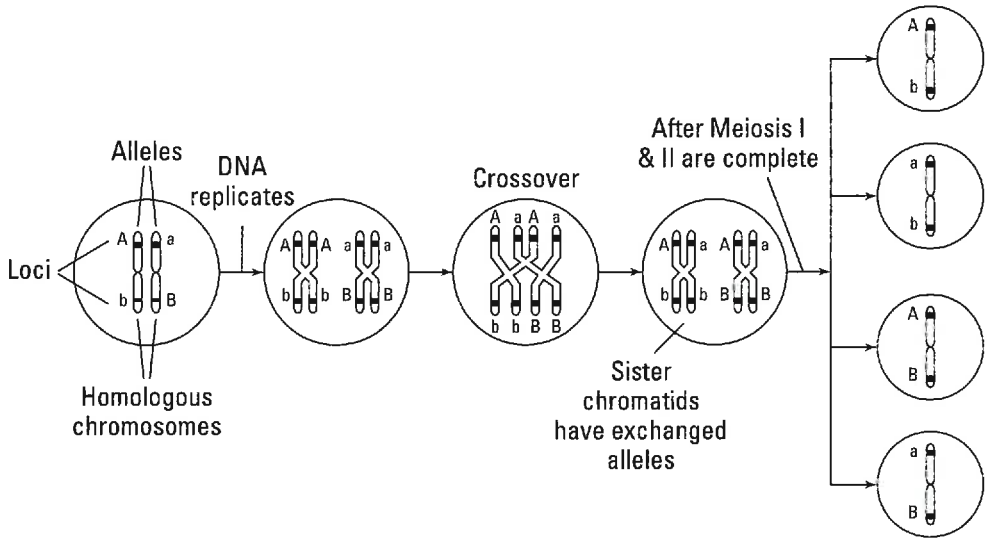


Figure 2-8: Crossing-over creates unique combinations of alleles during meiosis.

Meiosis II: The sequel

Meiosis II is the second phase of cell division that produces the final product of meiosis: cells that contain only one copy of each chromosome. The chromosomes condense once more to their now-familiar fat, sausage shapes. Keep in mind that each cell has only a single set of chromosomes, which are still in the form of sister chromatids.

During metaphase II, the chromosomes line up along the equator of the cells, and spindle fibers attach at the centromeres. In anaphase II, the sister chromatids are pulled apart and move to opposite poles of their respective cell. The nuclear membranes form around the now single chromosomes (telophase II). Finally, cell division takes place. At the end of the process, each of the four cells contains one single set of chromosomes.

Mommy, where did I come from?

From gametogenesis, honey. Meiosis in humans (and in all animals that reproduce sexually) produces cells called *gametes*. Gametes come in the form of sperm (produced by males) or eggs (produced by females). When conditions are right, sperm and egg unite to create a new organism, which takes the form of a *zygote*. Figure 2-9 shows the process of *gametogenesis* (the production of gametes) in humans.

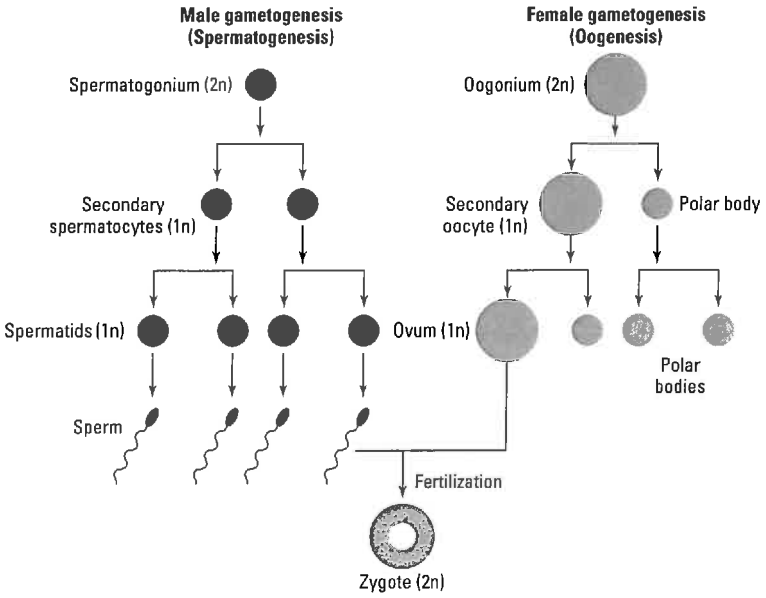


Figure 2-9: Gameto-genesis in humans.

For human males, special cells in the male's sexual organs (testes) produce *spermatogonia.* Spermatogonia are *2n* — they contain a full diploid set of 46 chromosomes (see the earlier section "Counting out chromosome numbers"). After meiosis I, each single spermatogonium has divided into two cells called *secondary spermatocytes.* These spermatocytes contain only one copy of each homolog (as sister chromatids). After one more division (meiosis II), the *spermatids* that become sperm cells have one copy of each chromosome. Thus, sperm cells are haploid and contain 23 chromosomes. Because males have X and Y sex chromosomes, half their sperm (men produce literally millions) contain Xs and half contain Ys.

Human females produce eggs in much the same way that men produce sperm. Egg cells, which are produced by the ovaries, start as diploid *oogonia* (that is, $2n = 46$). The big difference between egg and sperm production is that at the end of meiosis II, only one mature, haploid (23 chromosomes) sex cell (as an egg) is produced instead of four (refer to Figure 2-9). The other three cells produced are called *polar bodies;* the polar bodies aren't actual egg cells and can't be fertilized to produce offspring.

Why does the female body produce one egg cell and three polar bodies? Egg cells need large amounts of cytoplasm to nourish the zygote in the period between fertilization and when the mother starts providing the growing embryo with nutrients and energy through the placenta. The easiest way to get enough cytoplasm into the egg when it needs it most is to put less cytoplasm into the other three cells produced in meiosis II.