

# 4

## Evolution and Genetics

### CHAPTER OUTLINE

#### Evolution

Theory and Fact

#### Genetics

Mendel's Experiments

Independent Assortment and Recombination

#### Biochemical, or Molecular, Genetics

Cell Division

Crossing Over

Mutation

#### Population Genetics and Mechanisms of Genetic Evolution

Natural Selection

Random Genetic Drift

Gene Flow

#### The Modern Synthesis

Punctuated Equilibrium

### EVOLUTION

Compared with other animals, humans have uniquely varied ways—cultural and biological—of evolving, of adapting to environmental stresses. Exemplifying cultural adaptation, we manipulate our artifacts and behavior in response to environmental conditions. Contemporary North Americans turn up thermostats or travel to Florida in the winter. We turn on fire hydrants, swim, or ride in air-conditioned cars from New York City to Maine to escape the summer's heat. Although such reliance on culture has increased in the course of human evolution, people haven't stopped adapting biologically. As in other species, human populations adapt genetically in response to environmental forces, and individuals react physiologically to stresses. Thus, when we work in the midday sun, sweating occurs spontaneously, cooling the skin and reducing the temperature of subsurface blood vessels.



■ According to creationism, all life originated during the six days of Creation described in the Bible. Catastrophism proposed that fires and floods, including the biblical deluge involving Noah's ark (depicted in this painting by the American artist Edward Hicks), destroyed certain species. Note that creationism is not a scientific theory (see pp. 00–00, Chapter 1).

## OVERVIEW

Charles Darwin and Alfred Russel Wallace proposed that natural selection could explain the origin and evolution of species, as well as biological diversity among life forms. The *fact* of evolution was known prior to Darwin. The *theory* of evolution, through natural selection (*how* evolution occurred), was Darwin's major contribution. Darwin didn't know about the genetic mechanisms that allow natural selection to work. His contemporary Gregor Mendel made the pioneering discovery that genetic traits are inherited as discrete units, now called chromosomes and genes. Mendel also discovered that hereditary units may be inherited independently of each other and then reunite in new combinations, supplying some of the variety on which natural selection depends.

The adaptive value of particular traits depends on the environment. When the environment changes, selection works with traits that already are present in the population. If there isn't enough variety to permit adaptation to the environmental change, extinction is likely. Other evolutionary mechanisms work along with natural selection. Genetic drift operates most obviously in small populations, where pure chance can easily change gene frequencies. Gene flow and interbreeding keep subgroups of the same species connected genetically and thus work against speciation—the formation of new species.

We are ready now for a more detailed look at the principles that determine human biological adaptation, variation, and change.

During the 18th century, many scholars became interested in biological diversity, human origins, and our position within the classification of plants and animals. At that time, the commonly accepted explanation for the origin of species came from Genesis, the first book of the Bible: God had created all life during six days of Creation. According to **creationism**, biological similarities and differences originated at the Creation. Characteristics of life forms were seen as immutable; they could not change. Through calculations based on genealogies in the Bible, the biblical scholars James Ussher and John Lightfoot even claimed to trace the Creation to a very specific time: October 23, 4004 B.C. at 9 A.M.

Carolus Linnaeus (1707–1778) developed the first comprehensive and still influential classification, or taxonomy, of plants and animals. He grouped life forms on the basis of similarities and differences in their physical characteristics. He used traits such as the presence of a backbone to distinguish vertebrates from invertebrates and the presence of mammary glands to distinguish mammals from birds. Linnaeus viewed the differences between life forms as part of the Creator's orderly plan. Biological similarities and differences, he thought, had been established at the time of Creation and had not changed.

Fossil discoveries during the 18th and 19th centuries raised doubts about creationism. Fossils showed that different kinds of life had once existed. If all life had originated at the same time, why weren't ancient species still around? Why weren't contemporary plants and animals found in the fossil record? A modified explanation combining creationism with **catastrophism** arose to replace the original doctrine. In this view, fires, floods, and other catastrophes, including the biblical flood involving Noah's ark, had destroyed ancient species. After each destructive event, God had created again, leading to contemporary species. How did the catastrophists explain certain clear similarities between fossils and modern animals? They argued that some ancient species had managed to survive in isolated areas. For example, after the biblical flood, the progeny of the animals saved on Noah's ark spread throughout the world.

## Theory and Fact

The alternative to creationism and catastrophism was *transformism*, also called **evolution**. Evolutionists believe that species arise from others through a long and gradual process of transformation, or descent with modification. Charles Darwin became the best known of the evolutionists. However, he was influenced by earlier scholars, including his own grandfather. In a book

## The History of Chromosomes May Shape the Future of Diseases

NEW YORK TIMES NEWS BRIEF

by Carl Zimmer  
August 30, 2005

*The study of chromosomes, of which humans have 23 pairs, offers clues about evolutionary history and the origin of diseases—potentially leading to more effective treatment strategies. Described here is one category of mutation—chromosomal rearrangement—that can produce speciation, the formation of new species. This “News Brief” mentions two ways in which chromosomes are rearranged. One is inversion: A piece of the chromosome hives off, turns around, and is reattached. Another occurs when a piece splits off and migrates to a different part of the chromosome, where it reattaches. Such chromosomal rearrangements differ from single gene mutations that also are discussed in this chapter. As described in this news story, chromosomes have certain “hot spots” where breaks are most likely to occur, leading to rearrangements that sometimes influence speciation and at other times lead to diseases such as cancer.*

The common ancestor of humans and the rhesus macaque monkey lived about 25 million years ago. But despite that vast gulf of time, our chromosomes still retain plenty of evidence of our shared heritage.

A team of scientists at the National Cancer Institute recently documented this evidence by constructing a map of the rhesus macaque’s DNA, noting the location of 802 genetic markers in its genome. Then they compared the macaque map to a corresponding map of the human genome. The order of thousands of genes was the same.

“About half of the chromosomes are pretty much intact,” said William Murphy, a member of the team, now at Texas A&M University.

The other chromosomes had become rearranged over the past 25 million years, but Dr. Murphy and his colleagues were able to

reconstruct their evolution. Periodically, a chunk of chromosome was accidentally sliced out of the genome, flipped around and inserted backward.

In other cases, the chunk was ferried to a different part of the chromosome. All told, 23 of these transformations took place, and within these blocks of DNA, the order of the genes remained intact.

“It’s fairly easy to see how you can convert the chromosomes from the macaque to the human,” Dr. Murphy said.

This new macaque study, which is set to appear in a future issue of the journal *Genomics*, is just one of many new papers charting the history of chromosomes—in humans and other species. While scientists have been studying chromosomes for nearly a century, only in the last few years have large genome databases, powerful computers and new mathematical methods allowed scientists to trace these evolutionary steps.

Scientists hope that uncovering the history of chromosomes will have practical applications to diseases like cancer, in which rearranged chromosomes play a major part.

Scientists have known for over 70 years that chromosomes can be rearranged. With a microscope, it is possible to make out the banded patterns on chromosomes and to compare the pattern in different species.

Scientists discovered that different populations of fruit fly species could be distinguished by inverted segments in their chromosomes.

Later, molecular biologists discovered how cells accidentally rearranged large chunks of genetic material as they made new copies of their chromosomes. By the 1980’s, scientists were able to identify some major events in chromosome evolution. Humans have 23 pairs of chromosomes, for example, while chimpanzees and other apes have

24. Scientists determined that two ancestral chromosomes fused together after the ancestors of humans split off from other apes some six million years ago.

But a more detailed understanding of how chromosomes had changed would have to wait until scientists had amassed more information. In the mid-1990’s, Dr. Pevzner and Sridhar Hannenhalli of the University of Pennsylvania invented a fast method for comparing chromosomes from two different species and determining the fewest number of rearrangements . . . that separate them.

They introduced the method with a series of talks with titles like “Transforming Cabbage Into Turnips” and “Transforming Mice Into Men.” . . .

Dr. Pevzner himself joined with Dr. Murphy and 23 other scientists to analyze the last 100 million years of mammal evolution. They compared the genomes of humans to cats, dogs, mice, rats, pigs, cows and horses, using a program developed by Harris A. Lewin and his colleagues at the University of Illinois, called the Evolution Highway.

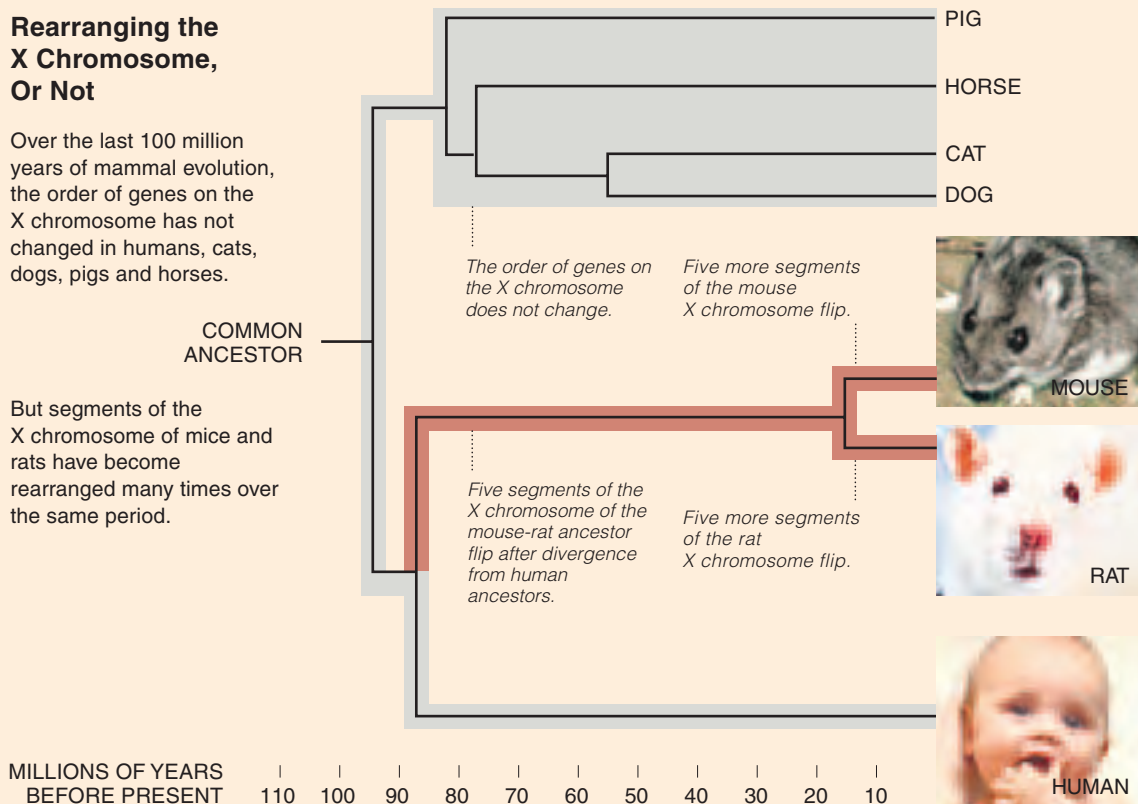
The program allowed them to trace how each lineage’s chromosomes had become rearranged over time. They published their results in the July 22 [2005] issue of *Science*.

The scientists found some chromosomes barely altered and others heavily reworked. They also discovered that the rate for rearrangements was far from steady. After the end of the Cretaceous Period, when large dinosaurs became extinct, the chromosomes of mammals began rearranging two to five times as fast as before. That may reflect the evolutionary explosion of mammals that followed the dinosaur extinctions, as mammals rapidly occupied new ecological niches as predators and grazers, fliers and swimmers . . .

## Rearranging the X Chromosome, Or Not

Over the last 100 million years of mammal evolution, the order of genes on the X chromosome has not changed in humans, cats, dogs, pigs and horses.

But segments of the X chromosome of mice and rats have become rearranged many times over the same period.



Sources: *Science*; William Murphy, Texas A&M University

The New York Times

The new results raise questions about how evolution makes chromosome rearrangements part of a species' genome. In many cases, these mutations cause diseases, so natural selection should make them disappear quickly from a population.

But scientists have also documented some rearrangements that are not hazardous or that are even beneficial. This year, for example, scientists discovered that some Northern Europeans carry a large inverted segment on one of their chromosomes. This inversion boosts the fertility of women who carry it. Chromosome rearrangements may also play a role in the origin of new species. Scientists often find that closely related species living in overlapping ranges have rearranged chromosomes. The mismatch of chromosomes may make it impossible for the two species to hybridize . . .

The *Science* study and the newer study on macaques suggest that

chromosomes tend to break in certain places, a hypothesis first offered by Dr. Pevzner in 2003 . . . "Certain regions of the genome are being broken over and over again."

It is too early to say why these regions have become break points, said Evan Eichler of the University of Washington, who was not involved in the mammal study. "There's something about these regions that makes them hot, and we have to figure out what that hot factor is," he said.

Dr. Eichler argues that it is important to figure out what that is because a number of human congenital diseases are associated with chromosome rearrangements at these same break points.

"Here you have a beautiful connection," he said. "The same thing that causes big-scale rearrangement between a human and chimp or a gorilla, these same sites are often the site of deletion associated with diseases."

Some of these diseases involve chromosome rearrangements in a fertilized egg, leading to congenital disorders. Cancer cells also undergo large-scale chromosome rearrangements, often at the same break points identified in the recent evolution study.

"We could have inherited some weaknesses in our genome that we have to understand and deal with medically," said David Haussler of the University of California, Santa Cruz. "And that has to do with the history of how our genome is built."

SOURCE: Carl Zimmer, "The History of Chromosomes May Shape the Future of Diseases," *New York Times*, August 30, 2005. <http://www.nytimes.com/2005/08/30/science/30gene.html?ei=5070&en=672b16463342c995&ex=1138165200&pagewanted=print>.

called *Zoonomia* published in 1794, Erasmus Darwin had proclaimed the common ancestry of all animal species.

Charles Darwin also was influenced by Sir Charles Lyell, the father of geology. During Darwin's famous voyage to South America aboard the *Beagle*, he read Lyell's influential book *Principles of Geology* (1837/1969), which exposed him to Lyell's principle of **uniformitarianism**. Uniformitarianism states that the present is the key to the past. Explanations for past events should be sought in the long-term action of ordinary forces that still operate today. Thus, natural forces (rainfall, soil deposition, earthquakes, and volcanic action) gradually have built and modified geological features such as mountain ranges. The earth's structure has been transformed gradually through natural forces operating for millions of years (see Weiner 1994).

Uniformitarianism was a necessary building block for evolutionary theory. It cast serious doubt on the belief that the world was only 6,000 years old. It would take much longer for such ordinary forces as rain and wind to produce major geological changes. The longer time span also allowed enough time for the biological changes that fossil discoveries were revealing. Darwin applied the ideas of uniformitarianism and long-term transformation to living things. He argued that all life forms are ultimately related and that the number of species has increased over time. (For more on science, evolution, and creationism, see Futuyma 1995; Gould 1999; Wilson 2002.)

Charles Darwin provided a theoretical framework for understanding evolution. He offered natural selection as a powerful evolutionary mechanism that could explain the origin of species, biological diversity, and similarities among related life forms. Darwin proposed a *theory of evolution* in the strict sense. A **theory** is a set of ideas formulated (by reasoning from known facts) to explain something. The main value of a



■ Charles Darwin at his home at Down House, Kent, England, around 1880.

theory is to promote new understanding. A theory suggests patterns, connections, and relationships that may be confirmed by new research. The *fact* of evolution (that evolution has occurred) was known earlier, for example by Erasmus Darwin. The *theory* of evolution, through natural selection (*how* evolution occurred), was Darwin's major contribution. Actually, natural selection wasn't Darwin's unique discovery. Working independently, the naturalist Alfred Russel Wallace had reached a similar conclusion (Shermer 2002). In a joint paper read to London's Linnaean Society in 1858, Darwin and Wallace made their discovery public. Darwin's book *On the Origin of Species* (1859/1958) offered much fuller documentation.

**Natural selection** is the process by which the forms most fit to survive and reproduce in a given environment do so in greater numbers than others in the same population. More than survival of the fittest, natural selection is differential reproductive success. Natural selection is a natural process that leads to a result. Natural selection operates when there is competition for strategic resources (those necessary for life) such as food and space between members of the population. There is also the matter of finding mates. You can win the competition for food and space and have no mate and thus have no impact on the future of the species. For natural selection to work on a particular population, there must be variety within that population, as there always is.

For information on common misconceptions about evolution, see your OLC Internet Exercises [mhhe.com/kottak](http://mhhe.com/kottak)



#### STUDENT CD-ROM LIVING ANTHROPOLOGY

Natural Selection

Track 4

This clip demonstrates the results of pigeon breeding in England, pointing out that more than 200 breeds of pigeons are known. The variety he observed among domestic birds (English pigeons) and wild birds (finches in the Galápagos Islands) led Charles Darwin to believe that comparable processes of selection were at work. In the first case the selection was artificial, the result of animal domestication and breeding experiments. In the second case the selection was natural, having to do with impersonal environmental forces. Are breeding experiments always useful to the animals in question? Which takes longer—artificial selection or natural selection?

## Intelligent Design versus Evolutionary Theory

“Intelligent design” (ID) no longer can be mentioned in biology classes in a Pennsylvania public school district, a federal district judge ruled on December 20, 2005. Dover Area School Board members had violated the Constitution when they ordered that its biology curriculum had to include the notion that life on earth was produced by an unspecified intelligent designer. Administrators had been required to read a statement in biology classes asserting that evolution was a theory, not a fact; that the evidence for evolution had gaps; and that ID offered an alternative explanation laid out in a book (purchased by church funds) in the school library. According to the judge (a Republican appointed by President George W. Bush), that statement amounted to an endorsement of religion. It could cause students to doubt a generally accepted scientific theory by presenting a religious alternative masquerading as a scientific theory (see *New York Times* 2005, p. A32).

The Dover school board policy, adopted in October 2004, was believed to have been the first of its kind in the United States. Their attorneys claimed that school board members were seeking to improve science education by exposing students to alternatives to Charles Darwin’s theory that evolution occurs through natural selection. ID proponents argued that evolutionary theory can’t fully explain complex life forms. Their opponents contended that ID amounts to a secular repackaging of creationism, which courts have ruled cannot be taught in public schools. The Pennsylvania judge agreed: The secular purposes claimed by the board were a pretext for the board’s real purpose—to promote religion in public schools.

ID advocates have since been voted off the Dover school board. The new board plans to remove ID from science classes, but interested students may be able to learn about ID in an elective course on comparative reli-

gion. ID did not belong in the science curriculum, the judge ruled, because it is “a religious view, a mere relabeling of creationism and not a scientific theory” (*New York Times* 2005, p. A32).

The ID movement asserts that life forms are too complex to have been formed by natural processes and must have been created by a higher intelligence. The fundamental claim of intelligent design proponents, such as William A. Dembski, is that “there are natural systems that cannot be adequately explained in terms of undirected natural forces and that exhibit features which in any other circumstance we would attribute to intelligence” (Dembski 2004). The source of this intelligence never is identified officially. But since the naturalness of the design is denied, its supernaturalness would seem to be assumed. By injecting ID into the science curriculum, the judge ruled, Dover’s board was unconstitutionally endorsing a religious view that advances “a particular version of Christianity” (*New York Times* 2005, p. A32). Attempts, of variable success and spurring ongoing legal challenges, have been made to teach ID in biology classes in several other states. ID’s greatest success has been in Kansas, where the State Board of Education has changed the definition of science so it is not limited to natural explanations. This opens the way for including ID and other forms of creationism.

The Pennsylvania court case thoroughly examined the claim that ID was science. After a six-week trial featuring hours of expert testimony, that claim was rejected. The judge found that ID violated the ground rules of science by invoking supernatural causation and by making assertions that could not be tested or proved wrong (falsified). Nor has ID gained acceptance in the scientific community. It lacks a research and testing program and is unsupported by peer-reviewed research (*New York Times* 2005).

Evolution as a *scientific theory* (as defined in the text) is a central orga-

nizing principle of modern biology and anthropology. Evolution also is a *fact*. There is absolutely no doubt that biological evolution has occurred and is occurring still. What is at issue in biology are questions of details of the process and the relative importance of different evolutionary mechanisms. “It is a *fact* that the earth with liquid water, is more than 3.6 billion years old. It is a *fact* that cellular life has been around for at least half of that period and that organized multicellular life is at least 800 million years old. It is a *fact* that major life forms now on earth were not at all represented in the past. There were no birds or mammals 250 million years ago. It is a *fact* that major life forms of the past are no longer living. There used to be dinosaurs . . . and there are none now. It is a *fact* that all living forms come from previous living forms. Therefore, all present forms of life arose from ancestral forms that were different. Birds arose from nonbirds and humans from nonhumans. No person who pretends to any understanding of the natural world can deny these facts any more than she or he can deny that the earth is round, rotates on its axis, and revolves around the sun” (Lewontin 1981, quoted in Moran 1993).

One key feature of science, as we saw in Chapter 1 (pp. 00–00), is to recognize the tentativeness and uncertainty of knowledge and understanding, which scientists try to improve. As they work to refine theories and to provide accurate explanations, scientists strive for objectivity and impartiality (trying to reduce the influence of the scientist, including his or her personal beliefs and actions). Science has many limitations and is not the only way we have of understanding. Certainly, the study of religion is another path to understanding. But the goals of objectivity and impartiality do help distinguish science from ways of knowing that are more biased, more rigid, and more dogmatic.

The giraffe's neck can illustrate how natural selection works on variety within a population. In any group of giraffes, there is always variation in neck length. When food is adequate, the animals have no problem feeding themselves. But when there is pressure on strategic resources, so that dietary foliage is not as abundant as usual, giraffes with longer necks have an advantage. They can feed off the higher branches. If this feeding advantage permits longer-necked giraffes to survive and reproduce even slightly more effectively than shorter-necked ones, giraffes with longer necks will transmit more of their genetic material to future generations than will giraffes with shorter necks.

An incorrect alternative to this (Darwinian) explanation would be the inheritance of acquired characteristics. That is the idea that in each generation, individual giraffes strain their necks to reach just a bit higher. This straining somehow modifies their genetic material. Over generations of strain, the average neck gradually gets longer through the accumulation of small increments of neck length acquired during the lifetime of each generation of giraffes. This is not how evolution works. If it did work in this way, weight lifters could expect to produce especially muscular babies. Workouts that promise no gain without the pain apply to the physical development of individuals, not species. Instead, evolution works as the process of natural selection takes advantage of the variety that is already present in a population. That's how giraffes got their necks.

Evolution through natural selection continues today. For example, in human populations there is differential resistance to disease, as we'll see in the discussion of sickle-cell anemia below. One classic recent example of natural selection is the peppered moth, which can be light or dark (in either case with black speckles, thus the name "peppered"). A change in this species illustrates recent natural selection (in our own industrial age) through what has been called *industrial melanism*. Great Britain's industrialization changed the environment to favor darker moths (those with more melanin) rather than the lighter-colored ones that were favored previously. During the 1800s industrial pollution increased; soot coated buildings and trees, turning them a darker color. The previously typical peppered moth, which had a light color, now stood out against the dark backgrounds of sooty buildings and trees. Such light-colored moths were easily visible to their predators. Through mutations (see below), a new strain of peppered moth, with a darker phenotype, was favored. Because these darker moths were fitter—that is harder to detect—in polluted environments, they survived and reproduced in greater numbers than lighter moths did. We see how natural selection may favor darker moths in polluted



■ Need photo caption.

environments and lighter-colored moths in non-industrial or less polluted environments because of their variant abilities to merge in with their environmental colors and thus avoid predators.

Evolutionary theory is used to explain. Remember from Chapter 1 that the goal of science is to increase understanding through explanation: showing how and why the thing (or class of things) to be understood (e.g., the variation within species, the geographic distribution of species, the fossil record) depends on other things. Explanations rely on associations and theories. An association is an observed relationship between two or more variables, such as the length of a giraffe's neck and the number of its offspring, or an increase in the frequency of dark moths as industrial pollution spreads. A theory is more general, suggesting or implying associations and attempting to explain them. A thing or event—for example, the giraffe's long neck—is explained if it illustrates a general principle or association, such as the concept of adaptive advantage. The truth of a scientific statement (e.g., evolution occurs because of differential reproductive success due to variation within the population) is confirmed by repeated observations. (See "Interesting Issues" for a discussion of differences between evolutionary theory and intelligent design.)

## GENETICS

Charles Darwin recognized that for natural selection to operate, there must be variety in the population undergoing selection. Documenting and explaining such variety among humans—human biological diversity—is one of anthropology's

■ Gregor Mendel,  
the father of genetics.



major concerns. Genetics, a science that emerged after Darwin, helps us understand the causes of biological variation. We now know that DNA (deoxyribonucleic acid) molecules make up genes and chromosomes, which are the basic hereditary units. Biochemical changes (mutations) in DNA provide much of the variety on which natural selection operates. Through sexual reproduction, recombination of the genetic traits of mother and father in each generation leads to new arrangements of the hereditary units received from each parent. Such genetic recombination also adds variety on which natural selection may operate.

**Mendelian genetics** studies the ways in which chromosomes transmit genes across the generations. **Biochemical genetics** examines structure, function, and changes in DNA. **Population genetics** investigates natural selection and other causes of genetic variation, stability, and change in breeding populations.

### Mendel's Experiments

In 1856, in a monastery garden, the Austrian monk Gregor Mendel began a series of experiments that were to reveal the basic principles of genetics. Mendel studied the inheritance of seven contrasting traits in pea plants. For each trait there were only two forms. For example, plants

were either tall (6 to 7 feet [1.8 to 2.1 meters]) or short (9 to 18 inches [23 to 46 centimeters]), with no intermediate forms. The ripe seeds could be either smooth and round or wrinkled. The peas could be either yellow or green, again with no intermediate colors.

When Mendel began his experiments, one of the prevailing beliefs about heredity was what has been called the "paint-pot" theory. According to this theory, the traits of the two parents blended in their children much as two pigments are blended in a can of paint. Children were therefore a unique mixture of their parents, and when these children married and reproduced, their traits would inextricably blend with those of their spouses. However, prevailing notions about heredity also recognized that occasionally the traits of one parent might swamp those of the other. If children looked far more like their mother than their father, people might say that her "blood" was stronger than his. Occasionally, too, there would be a "throwback," a child who was the image of his or her grandparent or who possessed a distinctive chin or nose characteristic of a whole line of descent.

Through his experiments with pea plants, Mendel discovered that heredity is determined by discrete particles or units. Although traits could disappear in one generation, they reemerged in their original form in later generations. For example, Mendel crossbred pure strains of tall and short plants. Their offspring were all tall. This was the first descending, or first filial, generation, designated  $F_1$ . Mendel then interbred the plants of the  $F_1$  generation to produce a generation of grandchildren, the  $F_2$  generation (Figure 4.1). In this generation, short plants reappeared. Among thousands of plants in the  $F_2$  generation, there was approximately one short plant for every three tall ones.

From similar results with the other six traits, Mendel concluded that although a **dominant** form could mask the other form in *hybrid*, or mixed, individuals, the dominated trait—the **recessive**—was not destroyed; it wasn't even changed. Recessive traits would appear in unaltered form in later generations because genetic traits were inherited as discrete units.

These basic genetic units that Mendel described were factors (now called genes or alleles) located on **chromosomes**. Chromosomes are arranged in matching (homologous) pairs. Humans have 46 chromosomes, arranged in 23 pairs, one in each pair from the father and the other from the mother.

For simplicity, a chromosome may be pictured as a surface (see Figure 4.2) with several positions, to each of which we assign a lowercase letter. Each position is a **gene**. Each gene determines, wholly or partially, a particular biological trait, such as whether one's blood is A, B, or O. **Alleles** (for example,  $b^1$  and  $b^2$  in Figure 4.2) are biochemically

different forms of a given gene. In humans, A, B, AB, and O blood types reflect different combinations of alleles of a particular gene.

In Mendel's experiments, the seven contrasting traits were determined by genes on seven different pairs of chromosomes. The gene for height occurred in one of the seven pairs. When Mendel crossbred pure tall and pure short plants to produce his F<sub>1</sub> generation, each of the offspring received an allele for tallness (T) from one parent and one for shortness (t) from the other. These offspring were mixed, or **heterozygous**, with respect to height; each had two dissimilar alleles of that gene. Their parents, in contrast, had been **homozygous**, possessing two identical alleles of that gene (see Hartl and Jones 2002).






















In the next generation (F<sub>2</sub>), after the mixed plants were interbred, short plants reappeared in the ratio of one short to three tall. Knowing that shorts only produced shorts, Mendel could assume that they were genetically pure. Another fourth of the F<sub>2</sub> plants produced only tall. The remaining half, like the F<sub>1</sub> generation, were heterozygous; when interbred, they produced three tall for each short. (See Figure 4.3.)

Dominance produces a distinction between **genotype**, or hereditary makeup, and **phenotype**, or expressed physical characteristics. Genotype is what you really are genetically; phenotype is what you appear as. Mendel's peas had three genotypes—TT, Tt, and tt—but only two phenotypes—tall and short. Because of dominance, the heterozygous plants were just as tall as the genetically pure tall ones. How do Mendel's discoveries apply to humans? Although some of our genetic traits follow Mendelian laws, with only two forms—dominant and recessive—other traits are determined differently. For instance, three alleles determine whether our blood type is A, B, AB, or O. People with two alleles for type O have that blood type. However, if they received a gene for either A or B from one parent and one for O from the other, they will have blood type A or B. In other words, A and B are both dominant over O. A and B are said to be *codominant*. If people inherit a gene for A from one parent and one for B from the other, they will have type AB blood, which is chemically different from the other varieties, A, B, and O.

These three alleles produce four phenotypes—A, B, AB, and O—and six different genotypes—OO, AO, BO, AA, BB, and AB (Figure 4.4). There are fewer phenotypes than genotypes because O is recessive to both A and B.

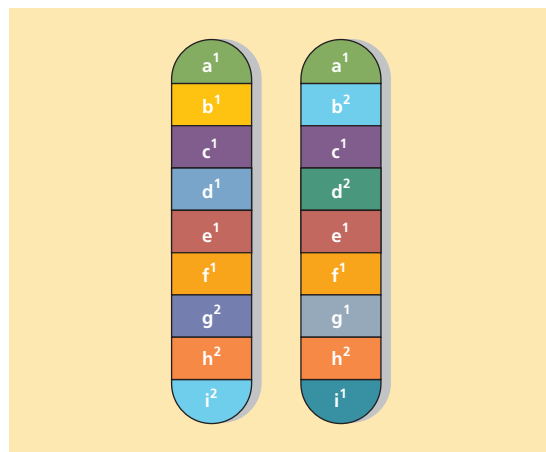
### Independent Assortment and Recombination

Through additional experiments, Mendel also formulated his law of **independent assortment**. He discovered that traits are inherited indepen-

| Trait Exhibited by F <sub>1</sub> Hybrids   | F <sub>2</sub> Generation (produced by crossbreeding F <sub>1</sub> hybrids)                         |  |
|---|--|--|
|   | Exhibit Dominant Trait   | Exhibit Recessive Trait  |
| Smooth seed shape<br>    | Smooth<br><br>3   | Wrinkled<br><br>1 |
| Yellow seed interior<br> | Yellow<br><br>3   | Green<br><br>1    |
| Gray seed coat<br>       | Gray<br><br>3     | White<br><br>1    |
| Inflated pod<br>         | Inflated<br><br>3 | Pinched<br><br>1  |
| Green pod<br>            | Green<br><br>3    | Yellow<br><br>1   |
| Axial pod<br>            | Axial<br><br>3    | Terminal<br><br>1 |
| Tall stem<br>           | Tall<br><br>3    | Short<br><br>1   |

Offspring exhibit dominant or recessive traits in ratio of 3:1.

**FIGURE 4.1** Mendel's Second Set of Experiments with Pea Plants. Dominant colors are shown unless otherwise indicated.



**FIGURE 4.2** Simplified Representation of a Normal Chromosome Pair. Letters indicate genes; superscripts indicate alleles.

|                  | F1    |    | F2    |    |
|------------------|-------|----|-------|----|
|                  | t     | t  | T     | t  |
| T                | Tt    | Tt | TT    | Tt |
| T                | Tt    | Tt | tT    | tt |
| Genotypic ratio  | 0:4:0 |    | 1:2:1 |    |
| Phenotypic ratio | 4:0   |    | 3:1   |    |

**FIGURE 4.3** Punnett Squares of a Homozygous Cross and a Heterozygous Cross.

These squares show how phenotypic ratios of the  $F_1$  and  $F_2$  generation are generated. Colors show genotypes.

|           |   | Parent I |        |       |
|-----------|---|----------|--------|-------|
|           |   | A        | B      | O     |
| Parent II | A | AA(A)    | AB(AB) | AO(A) |
|           | B | AB(AB)   | BB(B)  | BO(B) |
|           | O | AO(A)    | BO(B)  | OO(O) |

**FIGURE 4.4** Determinants of Phenotypes (Blood Groups) in the ABO System.

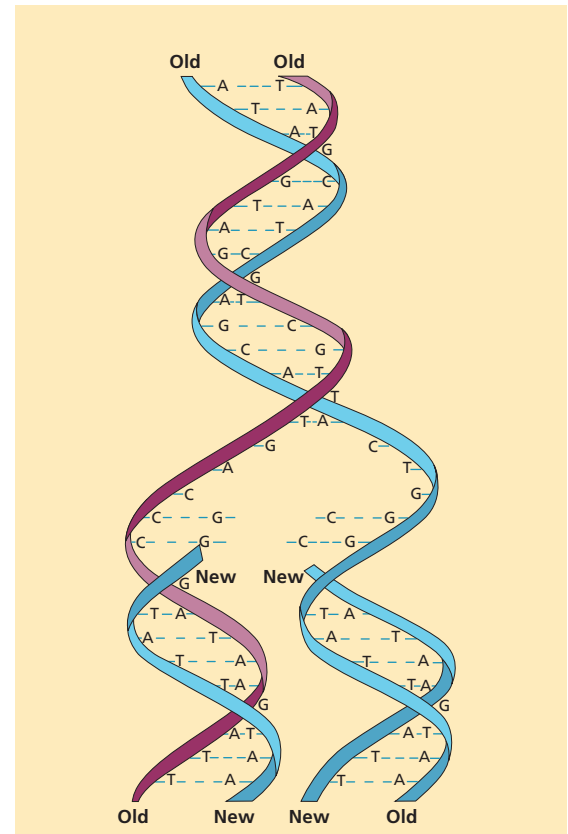
The four phenotypes—A, B, AB, and O—are indicated in parentheses and by color.

dently of one another. For example, he bred pure round yellow peas with pure wrinkled green ones. All the  $F_1$  generation peas were round and yellow, the dominant forms. But when Mendel interbred the  $F_1$  generation to produce the  $F_2$ , four phenotypes turned up. Round greens and wrinkled yellows had been added to the original round yellows and wrinkled greens.

The independent assortment and recombination of genetic traits provide one of the main ways by which variety is produced in any population. **Recombination** is important in biological evolution because it creates new types on which natural selection can operate.

## BIOCHEMICAL, OR MOLECULAR, GENETICS

If, as in Mendel's experiments, the same genetic traits always appeared in predictable ratios across the generations, there would be continuity rather than change. There would be no evolution. Various kinds of mutations produce the variety



**FIGURE 4.5** A double-stranded DNA molecule “unzips,” and a new strand forms on each of the old ones, producing two molecules, and eventually two cells, each identical to the first.

on which natural selection depends. Since Mendel's time, scientists have learned about **mutations**—changes in the DNA molecules of which genes and chromosomes are built. Mendel demonstrated that variety is produced by genetic recombination. Mutation, however, is even more important as a source of new biochemical forms on which natural selection may operate.

DNA does several things basic to life. DNA can copy itself, forming new cells, replacing old ones, and producing the sex cells, or *gametes*, that make new generations. DNA's chemical structure also guides the body's production of proteins—enzymes, antigens, antibodies, hormones, and hundreds of others.

The DNA molecule is a double helix (Crick 1962/1968; Watson 1970). Imagine it as a small rubber ladder that you can twist into a spiral. Its sides are held together by chemical bonds between four bases: thymine (T), adenine (A), cytosine (C), and guanine (G). DNA's duplication leads to ordinary cell division, as shown in Figure 4.5.

In protein building, another molecule, RNA, carries DNA's message from the cell's nucleus to its *cytoplasm* (outer area). The structure of RNA, with

paired bases, matches that of DNA. This permits RNA to carry a message from DNA in the cell nucleus to guide the construction of proteins in the cytoplasm. A protein, which is a chain of amino acids, is constructed by “reading” a length of RNA. RNA’s bases are read as three-letter “words,” called *triplets*—for example, AAG. (Because DNA and RNA have four bases, which can occur anywhere in the “word,” there are  $4 \times 4 \times 4 = 64$  possible triplets.) Each triplet “calls” a particular amino acid, although there is some redundancy; for example, AAA and AAG both call for the amino acid lysine. A protein is made as amino acids are assembled in the proper sequence.

Thus proteins are built following instructions sent by DNA, with RNA’s assistance. In this way, DNA, the basic *hereditary* material, also initiates and guides the construction of hundreds of proteins necessary for bodily growth, maintenance, and repair.

## Cell Division

An organism develops from a fertilized egg, or *zygote*, created by the union of two sex cells (gametes), a sperm from the father and an egg (ovum) from the mother. The zygote grows rapidly through **mitosis**, or ordinary cell division, which continues as the organism grows. Mistakes in this process of cell division, like the chromosomal breaks and rearrangements described in the news story, can cause diseases such as cancer.

The special process by which sex cells are produced is called **meiosis**. Unlike ordinary cell division, in which two cells emerge from one, in meiosis four cells are produced from one. Each has half the genetic material of the original cell. In human meiosis, four cells, each with 23 individual chromosomes, are produced from an original cell with 23 pairs.

With fertilization of egg by sperm, the father’s 23 chromosomes combine with the mother’s 23 to re-create the pairs in every generation. However, the chromosomes sort independently, so that a child’s genotype is a random combination of the DNA of its four grandparents. It is conceivable that one grandparent will contribute very little to its grandchild’s heredity. Independent assortment of chromosomes is a major source of variety, because the parents’ genotypes can be assorted in  $2^{23}$ , or more than 8 million, different ways.

## Crossing Over

Another source of variety is **crossing over**. Before fertilization, early in meiosis, as a sperm or egg is being formed, paired chromosomes temporarily intertwine as they duplicate themselves. As they do this, they often exchange lengths of their DNA (Figure 4.6). Crossovers are the sites where



■ The chromosomes that determine sex in humans. The X chromosome (left) is clearly larger than the Y chromosome (right). What are the genotypes of males and females in terms of these chromosomes?

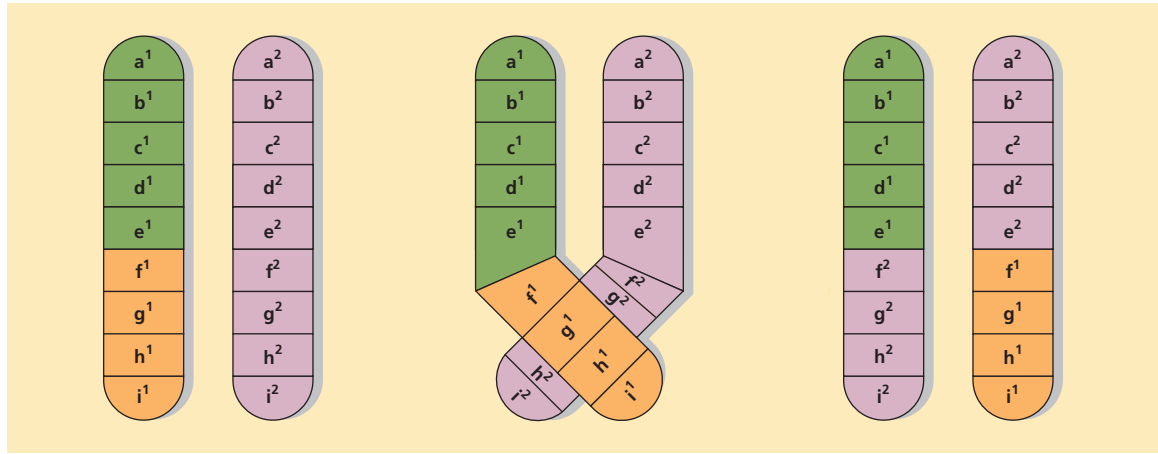
homologous chromosomes have exchanged segments by breakage and recombination.

Because of crossing over, each new chromosome is partially different from either member of the original pair. As a person produces sex cells, replacing, say, part of a chromosome one has received from one’s mother with a corresponding section of the homologous chromosome from one’s father, crossing over partially contradicts Mendel’s law of independent assortment and makes a new combination of genetic material available to the offspring. Because crossing over can occur with any chromosome pair, it is an important source of variety.

## Mutation

Mutations are the most important source of variety on which natural selection depends and operates. The simplest mutation results from substitution of just one base in a triplet by another. (This is called a *base substitution mutation*.) If such a mutation occurs in a sex cell that joins with another in a fertilized egg, the new organism will carry the mutation in every cell. As DNA directs protein building, a protein different from that produced by the nonmutant parent *may* be produced in the child. The child’s protein building will differ from the parent’s only if the new base codes for a different amino acid. Because the same amino acid can be coded by more than one triplet, a base substitution mutation doesn’t always produce a different protein. However, the abnormal protein associated with the hereditary disease sickle-cell anemia, described below, is caused by just such a difference in a single base between normal individuals and those afflicted with the disease.

Q: Check for sense.



**FIGURE 4.6** Crossing Over.

In the first phase of meiosis, homologous chromosomes intertwine as they duplicate themselves. As they do this, they often exchange lengths of their DNA, as shown here. This is known as crossing over. Note that the lower lengths of the original pair now differ. Each chromosome is therefore chemically different from either member of the original pair.

Another form of mutation—*chromosomal rearrangement*—was described in the “News Brief” at the beginning of the chapter. Pieces of a chromosome can break off, turn around and reattach, or migrate someplace else on that chromosome. This can occur in the sex cell, or in the fertilized egg or the growing organism, during mitosis. A mismatch of chromosomes resulting from rearrangement can lead to speciation (the formation of new species). Scientists often find that separate but closely related species living in overlapping ranges cannot interbreed because their chromosomes, due to rearrangement, no longer match. Chromosome rearrangements in a fertilized egg can lead to congenital disorders. Cancer cells undergo large-scale chromosome rearrangements. Chromosomes also may fuse. When the ancestors of humans split off from those of chimpanzees around six million years ago, two ancestral chromosomes fused together in the human line. Humans have 23 chromosome pairs, versus 24 for chimps.

Mutation rates vary, but for base substitution mutations, the likely average is  $10^{-9}$  mutations per DNA base per generation. This means that approximately three mutations will occur in every sex cell (Strachan and Read 2004). Many geneticists believe that most mutations are neutral, conferring neither advantage nor disadvantage. Others argue that most mutations are harmful and will be weeded out because they deviate from types that have been selected over the generations. However, if the selective forces affecting a population change, mutations in its gene pool may acquire an adaptive advantage they lacked in the old environment.

Evolution depends on mutations as a major source of genetically transmitted variety, raw

material on which natural selection can work. (Crossing over, independent assortment, and chromosomal recombination are other sources.) Alterations in genes and chromosomes may result in entirely new types of organisms, which may demonstrate some new selective advantage. Variants produced through mutation can be especially significant if there is a change in the environment. They may prove to have an advantage they lacked in the old environment. The spread of the allele that determines sickle-cell anemia, to be examined below, provides one example.

## POPULATION GENETICS AND MECHANISMS OF GENETIC EVOLUTION

Population genetics studies the stable and changing populations in which most breeding normally takes place (see Gillespie 2004; Hartl 2000). The term **gene pool** refers to all the alleles, genes, chromosomes, and genotypes within a breeding population—the “pool” of genetic material available. When population geneticists use the term *evolution*, they have a more specific definition in mind than the one given earlier (“descent with modification over the generations”). For geneticists, **genetic evolution** is defined as a change in gene frequency, that is, in the frequency of alleles in a breeding population from generation to generation. Any factor that contributes to such a change can be considered a mechanism of genetic evolution. Those mechanisms include natural selection, mutation (already examined), random genetic drift, and gene flow (see Mayr 2001).

## Natural Selection

Natural selection remains the best explanation for (genetic) evolution. Essential to understanding evolution through natural selection is the distinction between genotype and phenotype. Genotype refers just to hereditary factors—genes and chromosomes. Phenotype—the organism’s evident biological characteristics—develops over the years as the organism is influenced by particular environmental forces. (See the photo of the identical twins. Identical twins have exactly the same genotype, but their actual biology, their phenotypes, may differ as a result of variation in the environments in which they have been raised.) Also, because of dominance, individuals with different genotypes may have identical phenotypes (like Mendel’s tall pea plants). Natural selection can operate only on phenotype—on what is exposed, not on what is hidden. For example, a harmful recessive gene can’t be eliminated from the gene pool if it is masked by a favored dominant.

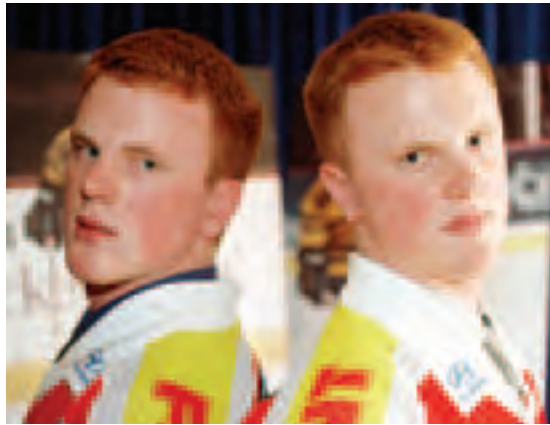
Phenotype includes not only outward physical appearance, but also internal organs, tissues, and cells and physiological processes and systems. Many biological reactions to foods, disease, heat, cold, sunlight, and other environmental factors are not automatic, genetically programmed responses but the product of years of exposure to particular environmental stresses. Human biology is not set at birth but has considerable *plasticity*. That is, it is changeable, being affected by the environmental forces, such as diet and altitude, that we experience as we grow up (see Bogin 2001).

The environment works on the genotype to build the phenotype, and certain phenotypes do better in some environments than other phenotypes do. However, remember that favored phenotypes can be produced by different genotypes. Because natural selection works only on genes that are expressed, maladaptive recessives can be removed only when they occur in homozygous form. When a heterozygote carries a maladaptive recessive, its effects are masked by the favored dominant. The process of perfecting the fit between organisms and their environment is gradual.

### Directional Selection

After several generations of selection, gene frequencies will change. Adaptation through natural selection will have occurred. Once that happens, those traits that have proved to be the most **adaptive** (favored by natural selection) in that environment will be selected again and again from generation to generation. Given such *directional selection*, or long-term selection of the same trait(s), maladaptive recessive alleles will be removed from the gene pool.

Directional selection will continue as long as environmental forces stay the same. However, if the environment changes, new selective forces



■ Need photo caption.

start working, favoring different phenotypes. This also happens when part of the population colonizes a new environment. Selection in the changed, or new, environment continues until a new equilibrium is reached. Then there is directional selection until another environmental change or migration takes place. Over millions of years, such a process of successive adaptation to a series of environments has led to biological modification and branching. The process of natural selection has led to the tremendous array of plant and animal forms found in the world today.

Selection operates *only* on traits that are present in a population. A favorable mutation *may* occur, but a population doesn’t normally come up with a new genotype or phenotype just because one is needed or desirable. Many species have become extinct because they weren’t sufficiently varied to adapt to environmental shifts.

There are also differences in the amount of environmental stress that organisms’ genetic potential enables them to tolerate. Some species are adapted to a narrow range of environments. They are especially endangered by environmental fluctuation. Others—*Homo sapiens* among them—tolerate much more environmental variation because their genetic potential permits many adaptive possibilities. Humans can adapt rapidly to changing conditions by modifying both biological responses and learned behavior. We don’t have to delay adaptation until a favorable mutation appears.

### Sexual Selection

Selection also operates through competition for mates in a breeding population. Males may openly compete for females, or females may choose to mate with particular males because they have desirable traits. Obviously, such traits vary from species to species. Familiar examples include color in birds; male birds, such as cardinals, tend to be more brightly colored than females are. Colorful males have a selective advantage because females

For an example of natural selection, see your OLC Internet Exercises

[mhhe.com/kottak](http://mhhe.com/kottak)



■ Need photo caption.

like them better. As, over the generations, females have opted for colorful mates, the alleles responsible for color have built up in the species. **Sexual selection**, based on differential success in mating, is the term for this process in which certain traits of one sex are selected because of advantages they confer in winning mates.

### Stabilizing Selection

We have seen that natural selection reduces variety in a population through directional selection—by favoring one trait or allele over another. Selective forces can also work to *maintain* variety through *stabilizing selection*, by favoring a **balanced polymorphism**, in which the frequencies of two or more alleles of a gene remain constant from generation to generation. This may be because the

phenotypes they produce are neutral, or equally favored, or equally opposed by selective forces. Sometimes a particular force favors (or opposes) one allele while a different but equally effective force favors (or opposes) the other allele.

One well-studied example of a balanced polymorphism involves two alleles,  $Hb^A$  and  $Hb^S$ , that affect the production of the beta strain (Hb) of human hemoglobin. Hemoglobin, which is located in our red blood cells, carries oxygen from the lungs to the rest of the body via the circulatory system. The allele that produces normal hemoglobin is  $Hb^A$ . Another allele,  $Hb^S$ , produces a different hemoglobin. Individuals who are homozygous for  $Hb^S$  suffer from *sickle-cell anemia*. Such anemia, in which the red blood cells are shaped like crescents or sickles, is associated with a disease that is usually fatal. This condition interferes with the blood's ability to store oxygen. It increases the heart's burden by clogging the small blood vessels.

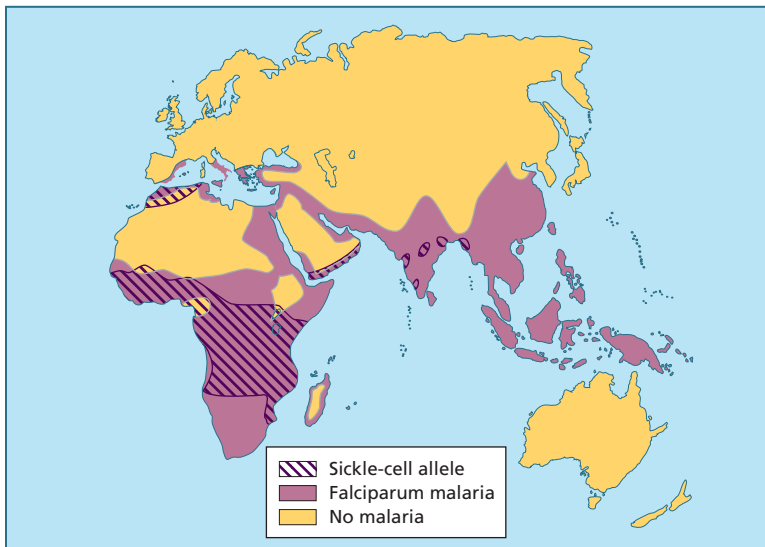
Given the fatal disease associated with  $Hb^S$ , geneticists were surprised to discover that certain populations in Africa, India, and the Mediterranean had very high frequencies of  $Hb^S$  (Figure 4.7). In some West African populations, that frequency is around 20 percent. Researchers eventually discovered that both  $Hb^A$  and  $Hb^S$  are maintained because selective forces in certain environments favor the heterozygote over either homozygote.

Initially, scientists wondered why, if most  $Hb^S$  homozygotes died before they reached reproductive age, the harmful allele hadn't been eliminated. Why was its frequency so high? The answer turned out to lie in the *heterozygote's* greater fitness. Only people who were homozygous for  $Hb^S$  died from sickle-cell anemia. Heterozygotes suffered very mild anemia, if any. On the other hand, although people homozygous for  $Hb^A$  did not suffer from anemia, they were much more susceptible to *malaria*—a killer disease that continues to plague *Homo sapiens* in the tropics.

The heterozygote, with one sickle-cell allele and one normal one, was the fittest phenotype for a malarial environment. Heterozygotes have enough abnormal hemoglobin, in which malaria parasites cannot thrive, to protect against malaria. They also have enough normal hemoglobin to fend off sickle-cell anemia. The  $Hb^S$  allele has been maintained in these populations because the heterozygotes survived and reproduced in greater numbers than did people with any other phenotype.

The example of the sickle-cell allele demonstrates the relativity of evolution through natural selection: Adaptation and fitness are in relation to specific environments. Traits are not adaptive or maladaptive for all times and places. Even harmful alleles can be selected if heterozygotes have an advantage. Moreover, as the environment

For an introduction to evolution, see the Virtual Exploration [mhhe.com/kottak](http://mhhe.com/kottak)



**FIGURE 4.7** Distribution of Sickle-Cell Allele and Falciparum Malaria in the Old World. SOURCE: Adapted from Joseph B. Birdsell, *Human Evolution: An Introduction to the New Physical Anthropology*, 3rd ed. (Boston: Houghton Mifflin, 1981).

## UNDERSTANDING OURSELVES

“Hey, it’s all in the genes.” When did you last hear a statement like that? We routinely use assumptions about genetic determination to explain, say, why tall parents have tall kids or why obesity runs in families. But how true is the statement? How much do our genes influence our bodies? The genetic causes of some of our physical traits are clear. This is true with the ABO blood group system, and with other blood factors, such as whether we are Rh positive or negative and whether we are sickle-cell carriers. But the genetic roots of other physical traits aren’t so clear. For example, can you crease or fold your tongue by raising its sides? Some people can; some people never can; some people who never thought they could can after practicing. An apparent genetic limitation turns out to be more plastic.

Human biology is plastic, but only to a degree. If you’re born with blood group O, you’ve got it for the rest of your life. The same applies to disorders due to harmful genes, such as those that cause hemophilia (transmitted on the X chromosome) and sickle-cell anemia. Still, if there’s no genetic solution, there may be a cultural one. Modern medicine now treats effectively a variety of genetic disorders that once would have been much more life threatening. Fortunately for us, plasticity through culture steps in to complement human biological plasticity.

changes, favored phenotypes and gene frequencies can change. In malaria-free environments, normal-hemoglobin homozygotes reproduce more effectively than heterozygotes do. With no malaria, the frequency of Hb<sup>S</sup> declines because Hb<sup>S</sup> homozygotes can’t compete in survival and reproduction with the other types. This has happened in areas of West Africa where malaria has been reduced through drainage programs and insecticides. Selection against Hb<sup>S</sup> also has occurred in the United States among Americans descended from West Africans (Diamond 1997).

### Random Genetic Drift

A second mechanism of genetic evolution is **random genetic drift**. This is a change in allele frequency that results not from natural selection but from chance. To understand why, compare the sorting of alleles to a game involving a bag of 12 marbles, 6 red and 6 blue. In step 1, you draw six marbles from the bag. Statistically, your chances of drawing three reds and three blues are less than



■ Beware the *Anopheles mosquito*, vector of malaria. An adult female is shown here.

those of getting four of one color and two of the other. Step 2 is to fill a new bag with 12 marbles on the basis of the ratio of marbles you drew in step 1. Assume that you drew four reds and two blues: The new bag will have eight red marbles and four blue ones. Step 3 is to draw six marbles from the new bag. Your chances of drawing blues in step 3 are lower than they were in step 1, and the probability of drawing all reds increases. If you do draw all reds, the next bag (step 4) will have only red marbles.

Although genetic drift can operate in any population, large or small, *fixation* due to drift is more rapid in small populations. Fixation refers to the total replacement of blue marbles by red marbles—or, to use a human example, of blue eyes by brown eyes. The history of the human line is characterized by a series of small populations, migrations, and fixation due to genetic drift. One cannot understand human origins, human genetic variation, and a host of other important anthropological topics without recognizing the importance of genetic drift.

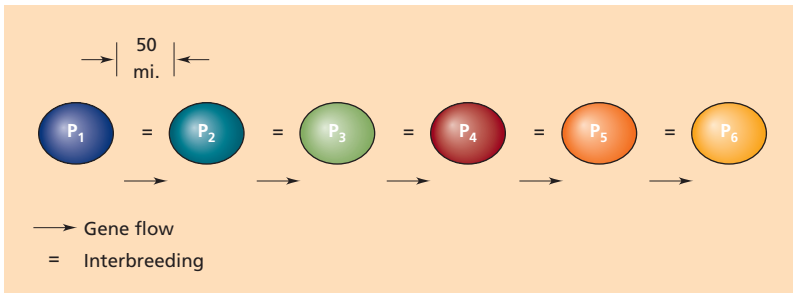
This game is analogous to random genetic drift operating over the generations. The blue marbles were lost purely by chance. Alleles, too, can be lost by chance rather than because of any disadvantage they confer. Lost alleles can reappear in a gene pool only through mutation.

### Gene Flow

A third mechanism of genetic evolution is **gene flow**, the exchange of genetic material between populations of the same species. Gene flow, like

### Biocultural Case Study

For more on the peopling of the Pacific and on how anthropology’s subfields unite to solve a particular problem, see the “Bringing It All Together” essay that immediately follows Chapter 11.



**FIGURE 4.8** Gene Flow between Local Populations.

$P_1$ – $P_6$  are six local populations of the same species. Each interbreeds (=) only with its neighbor(s). Although members of  $P_6$  never interbreed with  $P_1$ ,  $P_6$  and  $P_1$  are linked through gene flow. Genetic material that originates in  $P_1$  eventually will reach  $P_6$ , and vice versa, as it is passed from one neighboring population to the next. Because they share genetic material in this way,  $P_1$ – $P_6$  remain members of the same species. In many species, local populations distributed throughout a larger territory than the 250 miles depicted here are linked through gene flow.

mutation, works in conjunction with natural selection by providing variety on which selection can work. Gene flow may consist of direct interbreeding between formerly separated populations of the same species (e.g., Europeans, Africans, and Native Americans in the United States), or it may be indirect.

Consider the following hypothetical case (Figure 4.8). In a certain part of the world live six local populations of a certain species.  $P_1$  is the westernmost of these populations.  $P_2$ , which interbreeds with  $P_1$ , is located 50 miles to the east.  $P_2$  also interbreeds with  $P_3$ , located 50 miles east of  $P_2$ . Assume that each population interbreeds with, and only with, the adjacent populations.  $P_6$  is located 250 miles from  $P_1$  and does not directly interbreed with  $P_1$ , but it is tied to  $P_1$  through the chain of interbreeding that ultimately links all six populations.

Assume further that some allele exists in  $P_1$  that isn't particularly advantageous in its environment. Because of gene flow, this allele may be passed on to  $P_2$ , by it to  $P_3$ , and so on, until it eventually reaches  $P_6$ . In  $P_6$  or along the way, the allele may encounter an environment in which it does have a selective advantage. If this happens, it may serve, like a new mutation, as raw material on which natural selection can operate.

Alleles are spread through gene flow even when selection is not operating on the allele. In the long run, natural selection works on the variety within a population, whatever its source. Selection and gene flow have worked together to spread the  $Hb^S$  allele in Central Africa. Frequencies of  $Hb^S$  in Africa reflect not only the intensity of malaria but also the length of time gene flow has been going on (Livingstone 1969).

Gene flow is important in the study of the origin of species. A **species** is a group of related

organisms whose members can interbreed to produce offspring that can live and reproduce. A species has to be able to reproduce itself through time. We know that horses and donkeys belong to different species because their offspring cannot meet the test of long-term survival. A horse and a donkey may breed to produce a mule, but mules are sterile. So are the offspring of lions with tigers. Gene flow tends to prevent **speciation**—the formation of new species—unless subgroups of the same species are separated for a sufficient length of time.

When gene flow is interrupted, and isolated subgroups are maintained, new species may arise. Imagine that an environmental barrier arises between  $P_3$  and  $P_4$ , so that they no longer interbreed. If over time, as a result of isolation,  $P_1$ ,  $P_2$ , and  $P_3$  become incapable of interbreeding with the other three populations, speciation will have occurred.

## THE MODERN SYNTHESIS

The currently accepted view of evolution is known as the “modern synthesis.” This refers to the synthesis or combination of Darwin’s theory of evolution by natural selection and Mendel’s genetic discoveries. The modern synthesis also explains what Mendel could not—the inheritance of multifactorial or complex traits (e.g., height; see the next chapter). According to the modern synthesis, speciation (the formation of new species) occurs when they become reproductively isolated from one another. How does genetic evolution lead, or not, to new species?

**Microevolution** refers to genetic changes in a population or species over a few, several, or many generations, but without speciation. **Macroevolution** refers to larger-scale or more significant genetic changes in a population or species, usually over a longer time period, which result in speciation. Indeed, macroevolution is defined as speciation, the divergence of one ancestral species into two (or more) descendant species. Most biologists assume that species develop gradually as successive mutations accumulate in isolated populations, so that eventually the populations are too different to interbreed. But the time and the number of generations required for microevolution to become macroevolution are highly variable.

Modern-day creationists sometimes use a misunderstanding of the contrast between microevolution and macroevolution to comment on evolution. They may say they accept microevolution, such as a change in a species’ size or coloring, or as demonstrated in the laboratory or through studies of such traits as the sickle-cell allele. Macroevolution, they claim, by contrast, can’t be demonstrated, only inferred from the fossil record.



■ Tongue rolling—a genetic trait, at least partially. Some members of this family seem to be better at it than others are.

Note, however, that no degree of phenotypical difference is implied by the term *macroevolution*. In the “News Brief” at the beginning of this chapter, we saw that a simple chromosomal rearrangement can be sufficient to separate two closely related species whose ranges overlap. They belong to different species not because they are isolated from each other in space, but because they cannot hybridize. Although no phenotypic difference is visible between these reproductively isolated species, this is a case of macroevolution rather than microevolution.

To exaggerate the contrast between microevolution and macroevolution would imply, incorrectly, that there are two fundamentally distinct evolutionary processes. Scientists see no such contrast: Microevolution and macroevolution happen in the same way and for the same reasons, reflecting the mechanisms of genetic evolution discussed in this chapter. The modern synthesis recognizes that microevolutionary processes are sufficient to explain macroevolution.

### Punctuated Equilibrium

Charles Darwin saw species as arising from others over time, in a gradual and orderly fashion. Microevolutionary changes would accumulate over the generations to eventually produce macroevolution. In other words, minor alterations in the gene pool, accumulating generation after generation, would add up to major changes, including speciation, after thousands of years.

The **punctuated equilibrium** model of evolution (see Eldredge 1985; Gould 2002) points to the fact that long periods of stasis (stability), during

which species change little, may be interrupted (punctuated) by evolutionary leaps. One reason for such apparent jumps (which are revealed by the fossil record) may be extinction of one species followed by invasion by a closely related species. For example, a sea species may die out when a shallow body of water dries up, while a closely related species survives in deeper waters. Later, when the sea reinvades the first locale, the protected species will extend its range to the first area. Another possibility is that when barriers are removed, a group may replace, rather than succeed, a related one because it has a trait that makes it adaptively fitter in the environment they now share.

When there is a sudden environmental change, rather than such extinction and replacement, another possibility is for the pace of evolution to speed up. Some highly significant mutation(s) or combination of genetic changes may permit the survival of a radically altered species in a new and very different environmental niche. Many scientists believe that the evolution of our hominid ancestors was marked by one or more such evolutionary leaps.

Although species can survive radical environmental shifts, a more common fate is extinction. The earth has witnessed several mass extinctions—worldwide catastrophes affecting multiple species. The biggest one divided the era of “ancient life” (the Paleozoic) from the era of “middle life” (the Mesozoic). This mass extinction occurred 245 million years ago, when 4.5 million of the earth’s estimated 5 million species (mostly invertebrates) were wiped out. The second biggest extinction, around 65 million years ago, destroyed the dinosaurs. One

explanation for the extinction of the dinosaurs is that a massive, long-lasting cloud of gas and dust arose from the impact of a giant meteorite at the end of the Mesozoic. The cloud blocked solar radiation and therefore photosynthesis, ultimately destroying most plants and the chain of animals that fed on them.

From the fossil record, including the hominid fossil record to be discussed in Chapters 8 and 9, we know there are periods of more intense evolutionary change. The news story at the beginning

of this chapter describes variable mutation rates among species. At the end of the Mesozoic, the extinction of the dinosaurs was accompanied by the rapid spread and speciation of mammals and birds. Speciation responds to many factors, including the rate of environmental change, the speed with which geographic barriers rise or fall, the degree of competition with other species, and the effectiveness of the group's adaptive response. (See Appendix 1 for evolutionary theories applied to cultural change.)

## SUMMARY

1. In the 18th century, Carolus Linnaeus developed biological taxonomy. He viewed differences and similarities among organisms as part of God's orderly plan rather than as evidence for evolution. Charles Darwin and Alfred Russel Wallace proposed that natural selection could explain the origin of species, biological diversity, and similarities among related life forms. Natural selection requires variety in the population undergoing selection.
2. Through breeding experiments with peas in 1856, Gregor Mendel discovered that genetic traits pass on as units. These are now known to be chromosomes, which occur in homologous pairs. Alleles, some dominant, some recessive, are the chemically different forms that occur at a given genetic locus. Mendel also formulated the law of independent assortment. Each of the seven traits he studied in peas was inherited independently of all the others. Independent assortment of chromosomes and their recombination provide some of the variety needed for natural selection. But the major source of such variety is mutation, an alteration in the DNA molecules of which genes are made.
3. Biochemical, or molecular, genetics studies structure, function, and changes in genetic material—DNA. Genetic changes that provide variety within a population include base substitution mutations, chromosomal rearrangements, and genetic recombination. Population genetics studies gene frequencies in stable and changing populations. Natural selection is the most important mechanism of evolutionary change. Others include random genetic drift and gene flow. Natural selection works with traits already present in the population. If variety is insufficient to permit adaptation to environmental change, extinction is likely. New types don't appear just because they are needed.
4. One well-documented case of natural selection in contemporary human populations is that of the sickle-cell allele. In homozygous form, the sickle-cell allele,  $Hb^S$ , produces an abnormal hemoglobin. This clogs the small blood vessels, impairing the blood's capacity to store oxygen. The result is sickle-cell anemia, which is usually fatal. The distribution of  $Hb^S$  has been linked to that of malaria. Homozygotes for normal hemoglobin are susceptible to malaria and die in great numbers. Homozygotes for the sickle-cell allele die from anemia. Heterozygotes get only mild anemia and are resistant to malaria. In a malarial environment, the heterozygote has the advantage. This explains why an apparently maladaptive allele is preserved. The preservation of  $Hb^A$  and  $Hb^S$  alleles within a breeding population is an example of a balanced polymorphism, in which the heterozygote has greater fitness than does either homozygote.
5. Other mechanisms of genetic evolution complement natural selection. Random genetic drift operates most obviously in small populations, where pure chance can easily change allele frequencies. Gene flow and interbreeding keep subgroups of the same species genetically connected and thus impede speciation.
6. The modern synthetic theory of evolution (the modern synthesis) blends the Darwin and Wallace theory of evolution through natural selection with Mendel's discovery of the gene. Microevolution and macroevolution are two ends (short-term and long-term) of a continuum of evolutionary change in which gradually changing allele frequencies in a population eventually can lead to the formation of new species. Punctuated equilibrium theory states that long periods of stasis (stability), during which species change little, are interrupted (punctuated) by evolutionary leaps.

See the flash cards

[mhhe.com/kottak](http://mhhe.com/kottak)

**adaptive** Favored by natural selection in a particular environment.

**allele** A biochemical variant of a particular gene.

**balanced polymorphism** Two or more forms, such as alleles of the same gene, that maintain a constant frequency in a population from generation to generation.

**biochemical genetics** Field that studies structure, function, and changes in genetic material—aka molecular genetics.

**catastrophism** View that extinct species were destroyed by fires, floods, and other catastrophes. After each destructive event, God created again, leading to contemporary species.

**chromosomes** Basic genetic units, occurring in matching (homologous) pairs; lengths of DNA made up of multiple genes.

**creationism** Explanation for the origin of species given in Genesis: God created the species during the original six days of Creation.

**crossing over** During meiosis, the process by which homologous chromosomes intertwine and exchange segments of their DNA.

**dominant** Allele that masks another allele in a heterozygote.

**evolution** Belief that species arose from others through a long and gradual process of transformation, or descent with modification.

**gene** Area in a chromosome pair that determines, wholly or partially, a particular biological trait, such as whether one's blood type is A, B, or O.

**gene flow** Exchange of genetic material between populations of the same species through direct or indirect interbreeding.

**gene pool** All the alleles, genes, chromosomes, and genotypes within a breeding population—the “pool” of genetic material available.

**genetic evolution** Change in gene frequency within a breeding population.

**genotype** An organism's hereditary makeup.

**heterozygous** Having dissimilar alleles of a given gene.

**homozygous** Possessing identical alleles of a particular gene.

**independent assortment** Mendel's law of; chromosomes are inherited independently of one another.

**macroevolution** Large-scale changes in allele frequencies in a population over a longer time period (than microevolution)—changes that culminate in the evolution of new species.

**meiosis** Special process by which sex cells are produced; four cells are produced from one, each with half the genetic material of the original cell.

**Mendelian genetics** Studies ways in which chromosomes transmit genes across the generations.

**microevolution** Small-scale changes in allele frequencies over generations without speciation.

**mitosis** Ordinary cell division; DNA molecules copy themselves, creating two identical cells out of one.

**mutation** Change in the DNA molecules of which genes and chromosomes are built.

**natural selection** The process by which the forms most fit to survive and reproduce in a given environment do so in greater numbers than others in the same population; more than survival of the fittest, natural selection is differential reproductive success.

**phenotype** An organism's evident traits, its “manifest biology”—anatomy and physiology.

**population genetics** Field that studies causes of genetic variation, maintenance, and change in breeding populations.

**punctuated equilibrium** Evolutionary theory that long periods of stasis (stability), during which species change little, are interrupted (punctuated) by evolutionary leaps.

**random genetic drift** Change in gene frequency that results not from natural selection but from chance; most evident in small populations.

**recessive** Genetic trait masked by a dominant trait.

**recombination** Following independent assortment of chromosomes, new arrangements of hereditary units produced through bisexual reproduction.

**sexual selection** Based on differential success in mating, the process in which certain traits of one sex (e.g., color in male birds) are selected because of advantages they confer in winning mates.

**speciation** Formation of new species; occurs when subgroups of the same species are separated for a sufficient length of time.

**species** Population whose members can interbreed to produce offspring that can live and reproduce.

**theory** A set of ideas formulated (by reasoning from known facts) to explain something. The main value of a theory is to promote new understanding. A theory suggests patterns, connections, and relationships that may be confirmed by new research.

**uniformitarianism** Belief that explanations for past events should be sought in ordinary forces that continue to work today.

## CRITICAL THINKING QUESTIONS

1. If you are (or are pretending you are) a creationist, what do you see as the most convincing evidence for evolution?
2. If you are (or are pretending you are) an evolutionist, what do you see as the least convincing evidence for evolution?
3. Imagine that some of the seven traits that Mendel studied in pea plants were determined by genes on the same chromosome. How might his results have differed?
4. Is *Homo sapiens* more or less adaptable than other species? What makes us so adaptable? Can you think of some species that are more adaptable than we are?
5. Which of the mechanisms of genetic evolution acts to prevent speciation?

## SUGGESTED ADDITIONAL READINGS

- Cavalli-Sforza, L. L., P. Menozzi, and A. Piazza  
1994 *The History and Geography of Human Genes*. Princeton, NJ: Princeton University Press. Comprehensive look at the geographic spread of human genes.
- Cavalli-Sforza, L. L., and W. F. Bodmer  
1999 *The Genetics of Human Populations*. Mineola, NY: Dover. Principles and cases in population genetics, applied to humans.
- Eiseley, L.  
1961 *Darwin's Century*. Garden City, NY: Doubleday, Anchor Books. Discussion of Lyell, Darwin, Wallace, and other major contributors to natural selection and transformation.
- Futuyma, D. J.  
1995 *Science on Trial*, updated ed. Sunderland, MA: Sinauer Associates. The case of evolution versus creationism—favoring the former.  
1998 *Evolutionary Biology*. Sunderland, MA: Sinauer Associates. Basic text.
- Gillespie, J. H.  
2004 *Population Genetics: A Concise Guide*, 2nd ed. Baltimore: Johns Hopkins University Press. Good introduction to population genetics.
- Gould, S. J.  
1999 *Rock of Ages: Science and Religion in the Fullness of Life*. New York: Ballantine Books. Evolution, science, and religion by the well-known naturalist and science writer.  
2002 *The Structure of Evolutionary Theory*. Cambridge, MA: Belknap Press of Harvard University Press. Explores the punctuated equilibrium model and other aspects of evolutionary theory.
- Hartl, D. L.  
2000 *A Primer of Population Genetics*, 3rd ed. Sunderland, MA: Sinauer Associates. Short introduction to the field.
- Hartl, D. L., and E. W. Jones  
2002 *Essential Genetics*, 3rd ed. Sudbury, MA: Jones and Bartlett. Basic introduction to genetics.
- Lewontin, R.  
2000 *It Ain't Necessarily So: The Dream of the Human Genome and Other Illusions*. New York: New York Review of Books. Questions about nature, nurture, and contemporary genetic research.
- Mayr, E.  
2001 *What Evolution Is*. New York: Basic Books. A master scholar sums it all up.
- O'Rourke, D. H.  
2003 "Anthropological Genetics in the Genomic Era: A Look Back and Ahead." *American Anthropologist* 105(1):101–109. How the human genome project relates to anthropology.
- Shermer, M.  
2002 *In Darwin's Shadow: The Life and Science of Alfred Russel Wallace*. New York: Oxford University Press. The other inventor of natural selection.
- Weiner, J.  
1994 *The Beak of the Finch: A Story of Evolution in Our Time*. New York: Alfred A. Knopf. An excellent introduction to Darwin and to evolutionary theory.
- Wilson, D. S.  
2002 *Darwin's Cathedral: Evolution, Religion, and the Nature of Society*. Chicago: University of Chicago Press. Religion, sociology, and evolution.

1. Creationism: Look at the CreationWise cartoon website, <http://members.aol.com/dwr51055/humor.htm>. This site uses cartoons to express the concerns creationists have about evolution.
    - a. What is the creationist version of the origin of life? What evidence do creationists use to support their claims? According to these cartoons, what are some of the problems that creationists have with evolution?
    - b. What would be the response to these questions from a scientist who studies evolution?
  2. Human Blood Groups: Visit the following website of SCARF (Serum, Cells and Rare Fluid Exchange): <http://jove.prohosting.com/~scarfex/blood/groups.html>. The SCARF Exchange is an international group of scientists, physicians, and other individuals interested in human blood groups and transfusion medicine.
    - a. How many human blood group systems are listed? Click on ISBT number 004-RH. From what animal is the name RH derived? Do humans share the RH factor with that animal? What are the genetics of the RH factor? How can the RH factor affect fetal development? Where is the RH factor found, just in the blood or in other bodily fluids as well?
    - b. Click on ISBT number 001-ABO. Why is blood group testing important in giving and receiving blood? What are the genetics of the ABO system? On what chromosome are the ABO genes located? Where are the ABO antigens found, just in the blood or in other bodily fluids as well?
- See Chapter 4 at your McGraw-Hill Online Learning Center for additional review and interactive exercises.