1 Introduction to Genetics

Ansel Adams/National Park Archive at College Park, MD.

A Hopi pueblo on Black Mesa. Albinism, a genetic condition, arises with high frequency among the Hopi people and occupies a special place in the Hopi culture.

Albinism among the Hopis

Rising a thousand feet above the desert floor, Black Mesa dominates the horizon of the Enchanted Desert and provides a familiar landmark for travelers passing through northeastern Arizona. Black Mesa is not only a prominent geological feature: more significantly, it is the ancestral home of the Hopi Native Americans. Fingers of the mesa reach out into the desert, and alongside or on top of each finger is a Hopi village. Most of the villages are quite small, with only a few dozen inhabitants, but they are incredibly old. One village, Oraibi, has existed on Black Mesa since 1150 A.D. and is the oldest continually occupied settlement in North America.

In 1900, Alěs Hrdliěka, an anthropologist and physician working for the American Museum of Natural History, visited the Hopi villages of Black Mesa and reported a startling discovery. Among the Hopis were 11 white people—not Caucasians, but white Hopi Native Americans. These people had a genetic condition known as albinism (Figure 1.1).

1.1 Albinism among the Hopi Native Americans. The Hopi girl in the middle of this photograph, taken about 1900, has albinism.

Albinism is caused by a defect in one of the enzymes required to produce melanin, the pigment that darkens our skin, hair, and eyes. People with albinism either don't produce melanin or produce only small amounts of it, and consequently, have white hair, light skin, and no pigment in the irises of their eyes. Melanin normally protects the DNA of skin cells from the damaging effects of ultraviolet radiation in sunlight, and melanin's presence in the developing eye is essential for proper eyesight.

The genetic basis of albinism was first described by the English physician Archibald Garrod, who recognized in 1908 that the condition was inherited as an autosomal recessive trait, meaning that a person must receive two copies of an albino mutation—one from each parent—to have albinism. In recent years, the molecular natures of the mutations that lead to albinism have been elucidated. Albinism in humans is caused by defects in any one of several different genes that control the synthesis and storage of melanin. Many different types of mutations can occur in each gene, any one of which may lead to albinism. The form of albinism found among the Hopis is most likely oculocutaneous albinism (albinism affecting the eyes and skin) type 2, caused by a defect in the MA? 0 gene on chromosome 15.

The Hopis are not unique in having people with albinism among the members of their tribe. Albinism is found in almost all human ethnic groups and is described in ancient writings: it has probably been present since humankind's beginnings. What is unique about the Hopis is the high frequency of albinism in their population. In most human groups, albinism is rare, present in only about 1 in 20,000 people. In the villages on Black Mesa, it reaches a frequency of 1 in 200, a hundred times greater than in most other populations.

Why is albinism so frequent among the Hopis? The answer to this question is not completely known, but geneticists who have studied albinism among the Hopis speculate that the high frequency of the albino gene is at least partly related to the special place that albinism occupied in the Hopi culture. For much of their history, the Hopis considered members of their tribe with albinism to be important and special. People with albinism were considered pretty, clean, and intelligent. Having a number of people with albinism in one's village was considered a good sign, a symbol that the people of the village contained particularly pure Hopi blood. Members of the tribe with albinism performed in Hopi ceremonies and held positions of leadership, often as chiefs, healers, and religious leaders.

Hopis with albinism were also given special treatment in everyday activities. The Hopis have farmed small garden plots at the foot of Black Mesa for centuries. Every day throughout the growing season, the men of the tribe trek to the base of Black Mesa and spend much of the day in the bright southwestern sunlight tending their corn and vegetables. With little or no melanin in their skin, people with albinism are extremely susceptible to sunburn and have increased incidences of skin cancer when exposed to the sun. Furthermore, many don't see well in bright sunlight. Therefore, male Hopis with albinism were excused from farming and allowed to remain behind in the village with the women of the tribe, performing other duties.

Throughout the growing season, the men with albinism were the only male members of the tribe in the village with the women during the day, and thus they enjoyed a mating advantage, which helped to spread their albino genes. In addition, the special considerations given to Hopis with albinism allowed them to avoid the detrimental effects of albinism: increased skin cancer and

poor eyesight. The small size of the Hopi tribe probably also played a role by allowing chance to increase the frequency of the albino gene. Regardless of the factors that led to the high frequency of albinism, the Hopis greatly respected and valued the members of their tribe who possessed this particular trait. Unfortunately, people with genetic conditions in many societies are often subject to discrimination and prejudice. **TRY PROBLEMS 1 AND 22**

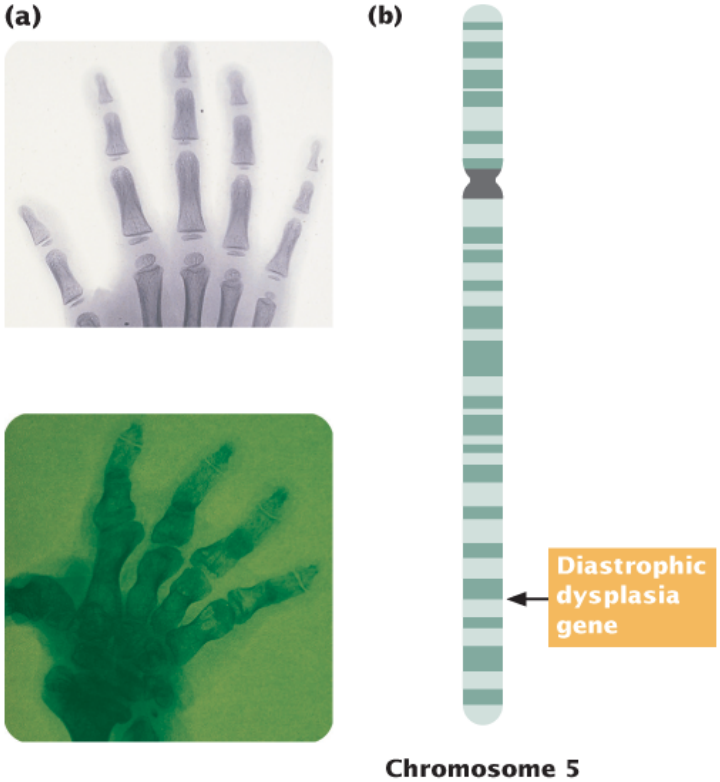
THINK-PAIR-SHARE Questions 1 and 2

Genetics is one of the most rapidly advancing fields of science, with important new discoveries reported every month. Look at almost any major news source and chances are that you will see articles related to genetics: the completion of another organism's genome, such as that of the monarch butterfly; the discovery of genes that affect major diseases, including multiple sclerosis, depression, and cancer; analyses of DNA from long-extinct animals such as the woolly mammoth; or the identification of genes that affect skin pigmentation, height, or learning ability in humans. Even among advertisements, you are likely to see ads for genetic testing to determine a person's ancestry, paternity, and susceptibility to diseases and disorders. These new findings and applications of genetics often have significant economic and ethical implications, making the study of genetics relevant, timely, and interesting.

This chapter introduces you to genetics and reviews some concepts that you may have encountered briefly in a preceding biology course. We begin by considering the importance of genetics to each of us, to society, and to students of biology. We then turn to the history of genetics and how the field as a whole developed. The final part of the chapter presents some fundamental terms and principles of genetics that are used throughout the book.

1.1 Genetics Is Important to Us Individually, to Society, and to the Study of Biology

Albinism among the Hopis illustrates the important role that genes play in our lives. This one genetic defect, among the 20,000 genes that humans possess, completely changes the life of a Hopi who possesses it. It alters his or her occupation, role in Hopi society, and relations with other members of the tribe. We all possess genes that influence our lives in significant ways. Genes affect our height, weight, hair color, and skin pigmentation. They influence our susceptibility to many diseases and disorders (**Figure 1.2**) and even contribute to our intelligence and personality. Genes are fundamental to who and what we are.

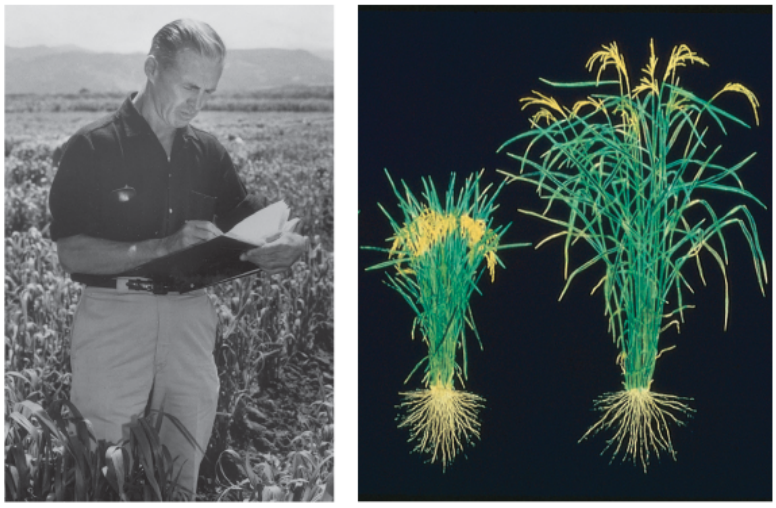


Part a: (top) Biophoto Associates/Science Source; (bottom) Reprinted from Cell, 78(6) Johanna Hästbacka, et al, The diastrophic dysplasia gene encodes a novel sulfate transporter: Positional cloning by fine-structure linkage disequilibrium mapping, pp. 1073 - 1087, ©1994 with permission from Elsevier. Permission conveyed through Copyright Clearance Center, Inc. Courtesy of Prof. Eric Lander, Whitehead Institute, MIT.

1.2 Genes influence susceptibility to many diseases and disorders. (a) An X-ray of the hand of a person suffering from diastrophic dysplasia (bottom), a hereditary growth disorder that results in curved bones, short limbs, and hand deformities, compared with an X-ray of a normal hand (top). (b) This disorder is due to a defect in the *SLC26A2* gene on chromosome 5.

Although the science of genetics is relatively new compared with sciences such as astronomy and chemistry, people have understood the hereditary nature of traits and have practiced genetics for thousands of years. The rise of agriculture began when people started to apply genetic principles to the domestication of plants and animals. Today, the major crops and animals used in agriculture are quite

different from their wild progenitors, having undergone extensive genetic alterations that increase their yields and provide many desirable traits, such as disease and pest resistance, special nutritional qualities, and characteristics that facilitate harvest. The Green Revolution, which expanded food production throughout the world in the 1950s and 1960s, relied heavily on the application of genetics (**Figure 1.3**). Today, genetically engineered corn, soybeans, and other crops constitute a significant proportion of the food produced worldwide.



Left: © Bettmann/CORBIS. Right: IRRI.

1.3 In the Green Revolution, genetic techniques were used to develop new high-yielding strains of crops. (Left) Norman Borlaug, a leader in the development of new varieties of wheat that led to the Green Revolution. Borlaug was awarded the Nobel Peace Prize in 1970. (Right) A modern, high-yielding rice plant (left) and a traditional rice plant (right).

The pharmaceutical industry is another area in which genetics plays an important role. Numerous drugs and food additives are synthesized by fungi and bacteria that have been genetically manipulated to make them efficient producers of these substances. The biotechnology industry employs molecular genetic techniques to develop and mass-produce substances of commercial value. Growth hormone, insulin, clotting factor, enzymes, antibiotics, vaccines, and many drugs are now produced commercially by genetically engineered bacteria and other cells (**Figure 1.4**). Genetics has also been used to produce bacteria that remove minerals from ore, break down toxic chemicals, and inhibit damaging frost formation on crop plants.

Reuters/Jerry Lampen

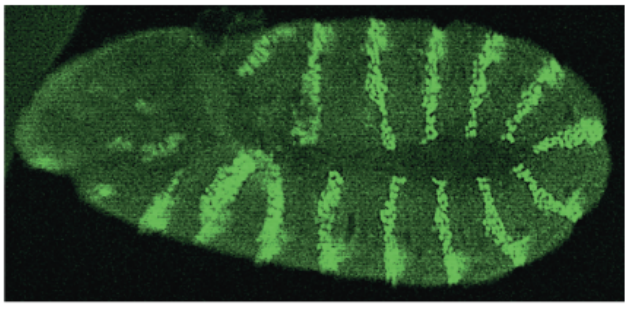
1.4 The biotechnology industry uses molecular genetic methods to produce substances of economic value.

Genetics also plays a critical role in medicine. Physicians recognize that many diseases and disorders have a hereditary component, including not only rare genetic disorders such as sickle-cell anemia and Huntington disease, but also many common diseases such as asthma, diabetes, and hypertension. Advances in genetics have resulted in important insights into the nature of diseases such as cancer and in the development of diagnostic tests, including those that identify pathogens and defective genes. Gene therapy—the direct alteration of genes to treat human diseases—has now been administered to thousands of patients, although its use is still experimental and limited.

THINK-PAIR-SHARE Question 3

The Role of Genetics in Biology

Although an understanding of genetics is important to all people, it is critical to the student of biology. Genetics provides one of biology's unifying principles: all organisms use genetic systems that have a number of features in common. Genetics also undergirds the study of many other biological disciplines. Evolution, for example, is genetic change that takes place over time, so the study of evolution requires an understanding of genetics. Developmental biology relies heavily on genetics: tissues and organs develop through the regulated expression of genes (**Figure 1.5**). Even such fields as taxonomy, ecology, and animal behavior are making increasing use of genetic methods. The study of almost any field of biology or medicine is incomplete without a thorough understanding of genes and genetic methods.



Steven Paddock.

1.5 The key to development lies in the regulation of gene expression. This early fruit-fly embryo illustrates

the localized expression (indicated by bright green) of the *engrailed* gene, which helps determine the development of body segments in the adult fly.

Genetic Diversity and Evolution

Life on Earth exists in a tremendous array of forms and features in almost every conceivable environment. Life is also characterized by adaptation: many organisms are exquisitely suited to the environment in which they are found. The history of life is a chronicle of new forms of life emerging, old forms disappearing, and existing forms changing.

Despite their tremendous diversity, living organisms have an important feature in common: all use similar genetic systems. The complete set of genetic instructions for any organism is its **genome**. All genomes are encoded in nucleic acids—either DNA or RNA. The coding system for genomic information is also common to all life: all genetic instructions are in the same format and, with rare exceptions, the code words are identical. Likewise, the processes by which genetic information is copied and decoded are remarkably similar for all forms of life. These common features of heredity suggest that all life on Earth evolved from the same primordial ancestor that arose between 3.5 billion and 4 billion years ago. Biologist Richard Dawkins describes life as a river of DNA that runs through time, connecting all organisms past and present.

genome
Complete set of genetic instructions for an organism.

The fact that all organisms have similar genetic systems means that the study of one organism's genes reveals principles that apply to other organisms. Investigations of how bacterial DNA is replicated (copied), for example, provide information that applies to the replication of human DNA. It also means that genes will function in foreign cells, which makes genetic engineering possible. Unfortunately, the similarity of genetic systems is also the basis for diseases such as AIDS (acquired immune deficiency syndrome), in which viral genes are able to function—sometimes with alarming efficiency—in human cells.

Life's diversity and adaptation are products of evolution, which is simply genetic change over time. Evolution is a two-step process: first, inherited differences arise randomly, and then the proportion of individuals with particular differences increases or decreases. Genetic variation is therefore the foundation of all evolutionary change and is ultimately the basis of all life as we know it. Furthermore, techniques of molecular genetics are now routinely used to decipher evolutionary relationships among organisms; for example, recent analysis of DNA isolated from Neanderthal fossils has provided insight into the relationship between Neanderthals and modern humans, demonstrating that Neanderthals and the ancestors of modern humans probably interbred some 30,000 to 40,000 years ago. Genetics, the study of genetic variation, is critical to understanding the past, present, and future of life.

THINK-PAIR-SHARE Question 4

CONCEPTS

Heredity affects many of our physical features as well as our susceptibility to many diseases and disorders. Genetics contributes to advances in agriculture, pharmaceuticals, and medicine and is fundamental to modern biology. All organisms use similar genetic systems, and genetic variation is the foundation of the diversity of all life.

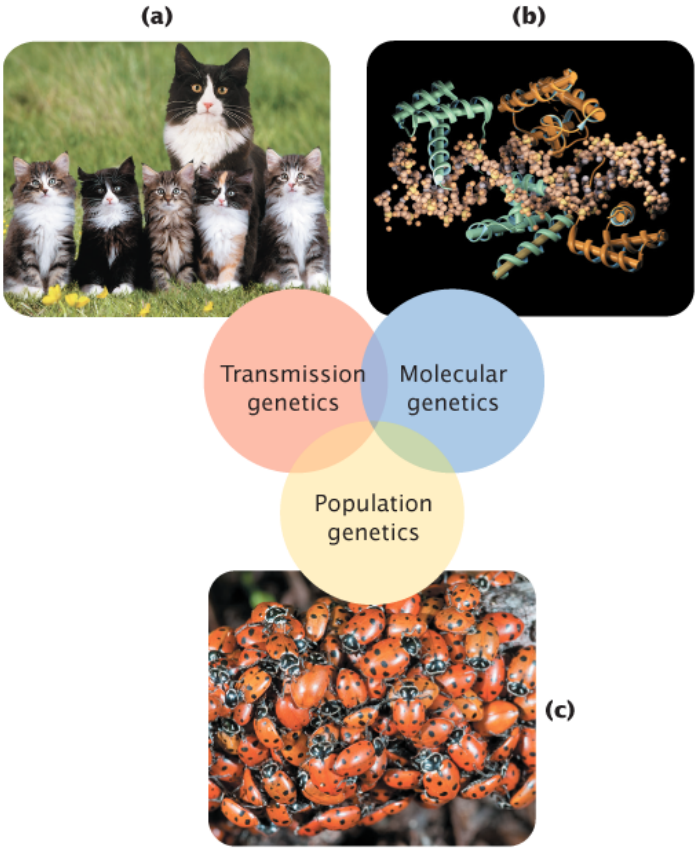
CONCEPT CHECK 1

What are some of the implications of all organisms having similar genetic systems?

a. That all life forms are genetically related
b. That research findings on one organism's gene function can often be applied to other organisms
c. That genes from one organism can often exist and thrive in another organism
d. All of the above

Divisions of Genetics

The study of genetics consists of three major subdisciplines: transmission genetics, molecular genetics, and population genetics (**Figure 1.6**). **Transmission genetics** (also known as classical genetics) encompasses the basic principles of heredity and how traits are passed from one generation to the next. This subdiscipline addresses the relation between chromosomes and heredity, the arrangement of genes on chromosomes, and gene mapping. Here, the focus is on the individual organism—how an individual inherits its genetic makeup and how it passes its genes to the next generation.



Top left: Juniors Bildarchiv/Alamy. Top right: Martin McCarthy/Getty Images.
Bottom: Stuart Wilson/Science Source.

1.6 Genetics can be subdivided into three interrelated fields.

transmission genetics
Field of genetics that encompasses the basic principles of genetics and how traits are inherited.

Molecular genetics concerns the chemical nature of the gene itself: how genetic information is encoded, replicated, and expressed. It includes the cellular processes of replication, transcription, and translation (by which genetic information is transferred from one molecule to another) and of gene

regulation (the processes that control the expression of genetic information). The focus in molecular genetics is the gene—its structure, organization, and function.

molecular genetics
Study of the chemical nature of genetic information and how it is encoded, replicated, and expressed.

Population genetics explores the genetic composition of groups of individuals of the same species (populations) and how that composition changes over time and space. Because evolution is genetic change, population genetics is fundamentally the study of evolution. The focus of this subdiscipline is the group of genes found in a population.

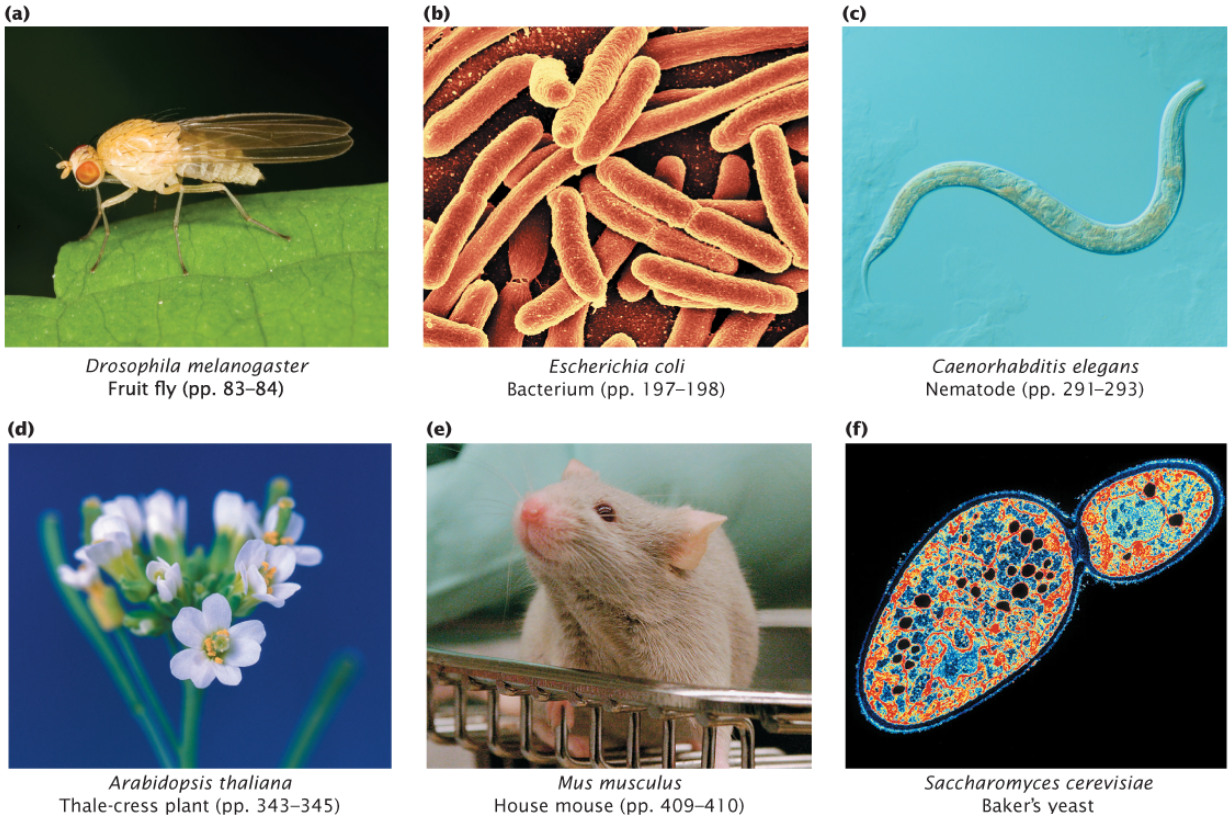
population genetics
Study of the genetic composition of populations (groups of members of the same species) and how their gene pools change with the passage of time.

The division of the study of genetics into these three subdisciplines is convenient and traditional, but we should recognize that these subdisciplines overlap and that each one can be further divided into a number of more specialized fields, such as chromosomal genetics, biochemical genetics, quantitative genetics, and so forth. Alternatively, the study of genetics can be subdivided by organism (fruit-fly, corn, or bacterial genetics), and each of these organisms can be studied at the level of transmission, molecular, or population genetics. Modern genetics is an extremely broad field, encompassing many interrelated subdisciplines and specializations. ▶ **TRY PROBLEMS 17**

Model Genetic Organisms

Through the years, genetic studies have been conducted on thousands of different species, including almost all major groups of bacteria, fungi, protists, plants, and animals. Nevertheless, a few species have emerged as **model genetic organisms**: organisms with characteristics that make them particularly useful for genetic analysis and about which a tremendous amount of genetic information has accumulated. Six model organisms that have been the subject of intensive genetic study are *Drosophila melanogaster*, a fruit fly; *Escherichia coli*, a bacterium present in the gut of humans and other mammals; *Caenorhabditis elegans*, a nematode (also called a roundworm); *Arabidopsis thaliana*, the thale cress plant; *Mus musculus*, the house mouse; and *Saccharomyces cerevisiae*, baker's yeast (**Figure 1.7**). These species are the organisms of choice for many genetic researchers, and their genomes were sequenced as a part of the Human Genome Project (described in Chapter 15).

model genetic organism
An organism that is widely used in genetic studies because it has characteristics, such as short generation time and large numbers of progeny, that make it well suited to genetic analysis.

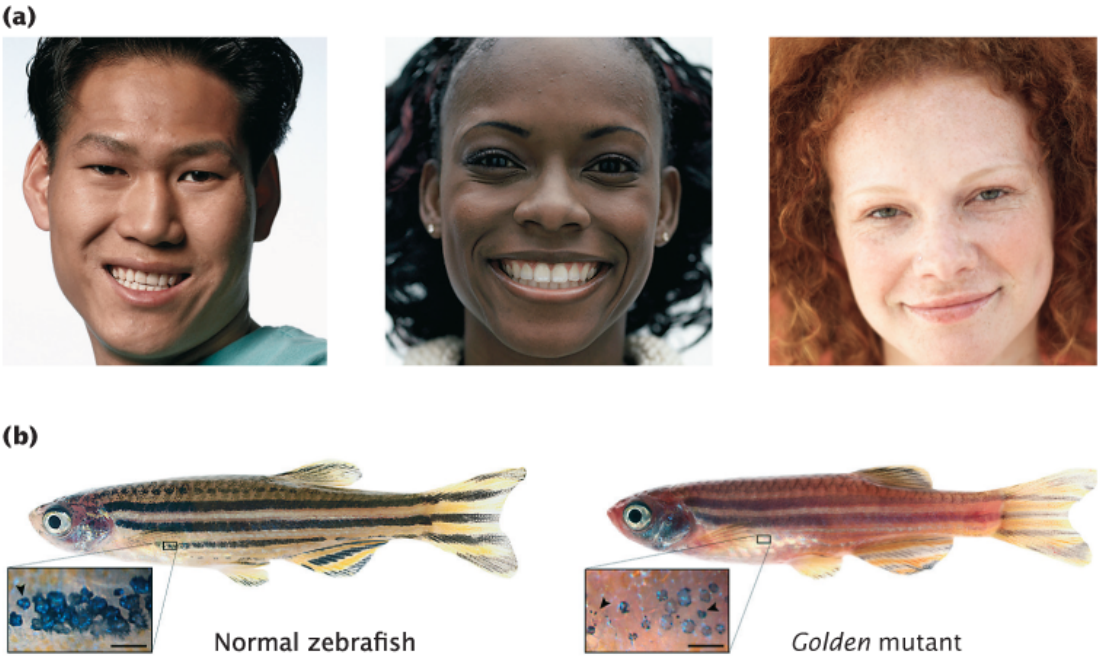


Part a: Alfred Schauhuber / imageBROKER / Alamy Stock Photo. Part b: Pasieka/Science Source.
Part c: Sinclair Stammers/Science Source. Part d: Peggy Greb/ARS/USDA.
Part e: AP Photo/Joel Page. Part f: Biophoto Associates/Science Source.

1.7 Model genetic organisms are species with features that make them useful for genetic analysis. Organisms (a) through (e) are discussed in more detail on the pages referred to below each.

At first glance, this group of lowly and sometimes unappreciated creatures might seem to be unlikely candidates for model organisms. However, all possess traits that make them particularly suitable for genetic study, including a short generation time, large but manageable numbers of progeny, adaptability to a laboratory environment, and the ability to be housed and propagated inexpensively. The life cycles, genomic characteristics, and features that make these model organisms useful for genetic studies are included in special illustrations in later chapters for five of the six species. Other species that are frequently the subject of genetic research and considered model genetic organisms include *Neurospora crassa* (bread mold), *Zea mays* (corn), *Danio rerio* (zebrafish), and *Xenopus laevis* (clawed frog). Although not generally considered a model genetic organism, humans have also been subjected to intensive genetic scrutiny.

The value of model genetic organisms is illustrated by the use of zebrafish to identify genes that affect skin pigmentation in humans. For many years, geneticists have recognized that differences in pigmentation among human ethnic groups (**Figure 1.8a**) are genetic, but the genes causing these differences were largely unknown. The zebrafish has become an important model in genetic studies because it is a small vertebrate that produces many offspring and is easy to rear in the laboratory. The mutant zebrafish called *golden* has light pigmentation due to the presence of fewer, smaller, and less dense pigment-containing structures called melanosomes in its cells (**Figure 1.8b**). Light skin in humans is similarly due to fewer and less dense melanosomes in pigment-containing cells.



Part a: Barbara Penoyar/Getty Images; Amos Morgan/Getty Images; Stockbyte/Getty Images. Part b: Keith Cheng/Jake Gittlen, Cancer Research Foundation Penn State College of Medicine.

1.8 The zebrafish, a model genetic organism, has been instrumental in helping to identify genes encoding pigmentation differences among humans. (a) Human ethnic groups differ in degree of skin pigmentation. (b) The zebrafish *golden* mutation is caused by a gene that controls the amount of melanin in melanosomes.

Keith Cheng and his colleagues hypothesized that light skin in humans might result from a mutation that is similar to the *golden* mutation in zebrafish. Taking advantage of the ease with which zebrafish can be manipulated in the laboratory, they isolated and sequenced the gene responsible for the *golden* mutation and found that it encodes a protein that takes part in calcium uptake by melanosomes. They then searched a database of all known human genes and found a similar gene called *SLC24A5*, which encodes the same function in human cells. When they examined human populations, they found that light-skinned Europeans typically possess one form of this gene, whereas darker-skinned Africans, East Asians, and Native Americans usually possess a different form of the gene. Many other genes also affect pigmentation in humans, as illustrated by mutations in the *OCA2* gene that produce albinism among the Hopis (discussed in the introduction to this chapter). Nevertheless, *SLC24A5* appears to be responsible for 24% to 38% of the differences in pigmentation between Africans and Europeans. This example illustrates the power of model organisms in genetic research. However, we should not forget that all organisms possess unique characteristics and that sometimes the genetics of models do not accurately reflect the genetic systems of other organisms.

CONCEPTS

The three major divisions of genetics are transmission genetics, molecular genetics, and population genetics. Transmission genetics examines the principles of heredity; molecular genetics deals with the gene and the cellular processes by which genetic information is transferred and expressed; population genetics concerns the genetic composition of groups of organisms and how that composition changes over time and space. Model genetic organisms are species that have received special emphasis in genetic research: they have characteristics that make them useful for genetic analysis.

CONCEPT CHECK 7

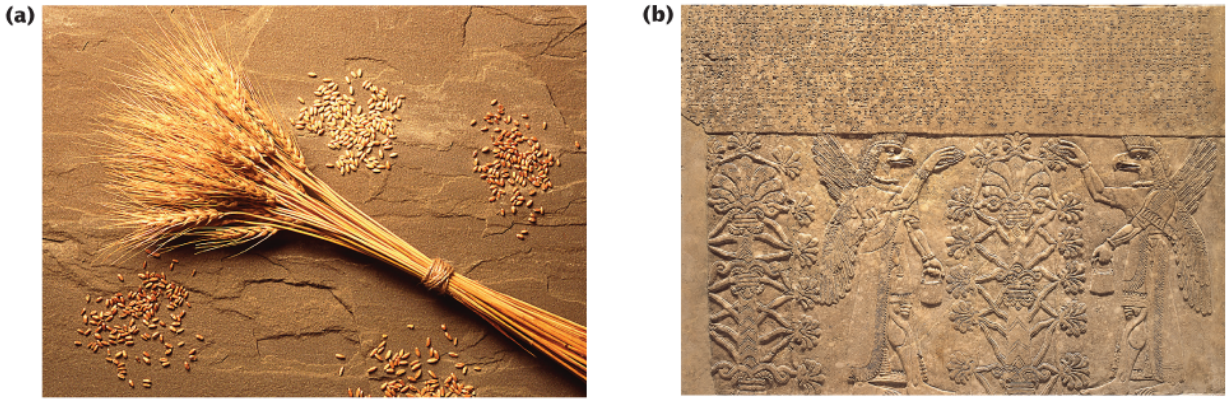
Would the horse make a good model genetic organism? Why or why not?

1.2 Humans Have Been Using Genetics for Thousands of Years

Although the science of genetics is young—almost entirely a product of the past 100 years or so—people have been using genetic principles for thousands of years.

The Early Use and Understanding of Heredity

The first evidence that people understood and applied the principles of heredity in earlier times is found in the domestication of plants and animals, which began between approximately 10,000 and 12,000 years ago in the Middle East. The first domesticated organisms included wheat, peas, lentils, barley, dogs, goats, and sheep (Figure 1.9a). By 4000 years ago, sophisticated genetic techniques were already in use in the Middle East. The Assyrians and Babylonians developed several hundred varieties of date palms that differed in fruit size, color, taste, and time of ripening (Figure 1.9b). Other crops and domesticated animals were developed by cultures in Asia, Africa, and the Americas in the same period.



Part a: Scott Bauer/ARS/USDA. Part b: Image copyright © The Metropolitan Museum of Art. Image source: Art Resource, NY.

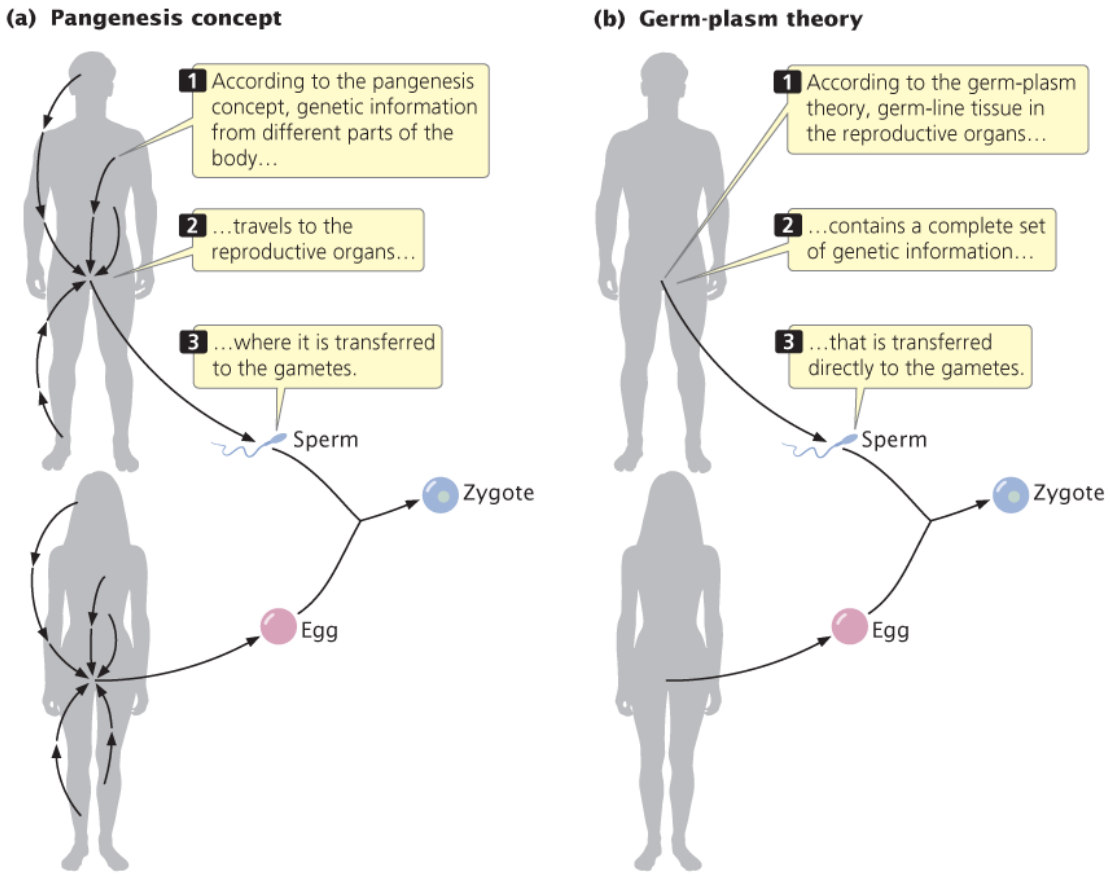
1.9 Ancient peoples practiced genetic techniques in agriculture. (a) Modern wheat, with larger and more numerous seeds that do not scatter before harvest, was produced by interbreeding at least three different wild species. (b) Assyrian bas-relief sculpture showing artificial pollination of date palms at the time of King Assurnasirpalli II, who reigned from 883 to 859 B.C.

The ancient Greeks gave careful consideration to human reproduction and heredity. Greek philosophers developed the concept of **pangenesis**. This concept suggested that specific pieces of information travel from various parts of the body to the reproductive organs, from which they are passed to the embryo (Figure 1.10a). Pangenesis led the ancient Greeks to propose the notion of the **inheritance of acquired characteristics**, in which traits acquired in a person's lifetime become incorporated into that person's hereditary information and are passed on to offspring; for example, people who developed musical ability through diligent study would produce children who are innately endowed with musical ability. Although incorrect, these ideas persisted through the twentieth century.

pangenesis
Early concept of heredity proposing that particles carry genetic information from different parts of the body to the reproductive organs.

inheritance of acquired characteristics

Early notion of inheritance proposing that acquired traits are passed to descendants.

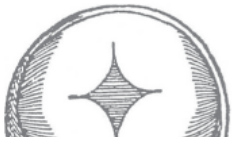


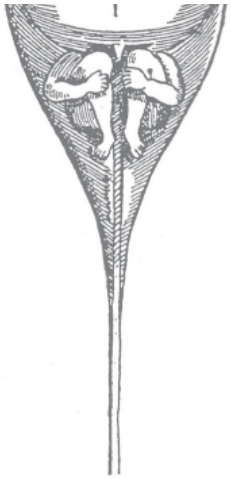
Pierce, *Genetics Essentials: Concepts and Connections*, 4e, © 2018 W. H. Freeman and Company

1.10 Pangenesis, an early concept of inheritance, compared with the modern germ-plasm theory.

Additional developments in our understanding of heredity occurred during the seventeenth century. Dutch eyeglass makers began to put together simple microscopes in the late 1500s, enabling Robert Hooke (1635–1703) to discover cells in 1665. Microscopes provided naturalists with new and exciting vistas on life, and perhaps it was excessive enthusiasm for this new world of the very small that gave rise to the idea of **preformationism**. According to preformationism, inside the egg or sperm there exists a fully formed miniature adult, a fmksl asjsq which simply enlarges during development (**Figure 1.11**). Preformationism meant that all traits were inherited from only one parent—from the father if the homunculus was in the sperm or from the mother if it was in the egg. Although many observations suggested that offspring possess a mixture of traits from both parents, preformationism remained a popular concept throughout much of the seventeenth and eighteenth centuries.

preformationism
Early concept of inheritance proposing that a miniature adult (homunculus) resides in either the egg or the sperm and increases in size during development, with all traits inherited from the parent that contributes the homunculus.





Science Source.

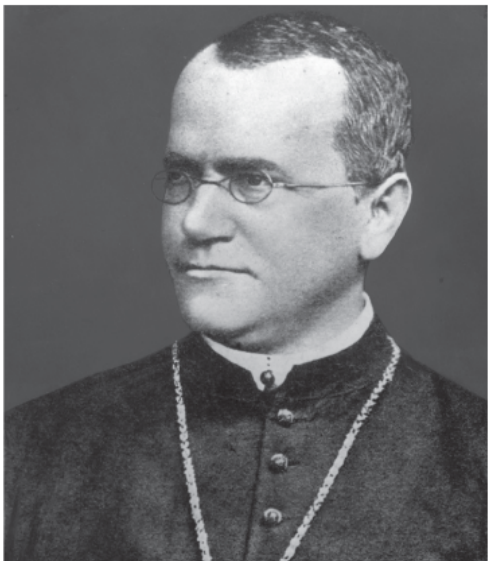
1.11 Preformationists in the seventeenth and eighteenth centuries believed that a sperm or an egg contains a fully formed human (the homunculus). Shown here is a drawing of a homunculus inside a sperm.

Another early notion of heredity was **blending inheritance**, which proposed that the traits of offspring are a blend, or mixture, of parental traits. This idea suggested that the genetic material itself blends, much as blue and yellow pigments blend to make green paint. It also suggested that after having been blended, genetic differences could not be separated in future generations, just as green paint cannot be separated into blue and yellow pigments. Some traits do _nnc_pto exhibit blending inheritance; however, thanks to Gregor Mendel's research with pea plants, we now understand that individual genes do not blend.

blending inheritance
Early concept of heredity proposing that offspring possess a mixture of the traits from both parents.

The Rise of the Science of Genetics

In 1676, Nehemiah Grew (1641–1712) reported that plants reproduce sexually by using pollen from the male sex cells. With this information, a number of botanists, including Gregor Mendel (1822–1884; **Figure 1.12**), began to experiment with crossing plants and creating hybrids. Mendel went on to discover the basic principles of heredity in the 1860s.



Hulton Archive/Getty Images.

1.12 Gregor Mendel was the founder of modern genetics. Mendel first discovered the principles of heredity by crossing different varieties of pea plants and analyzing the pattern of the transmission of traits in subsequent generations.

Developments in cytology (the study of cells) in the 1800s had a strong influence on genetics. Building on the work of others, Matthias Jacob Schleiden (1804–1881) and Theodor Schwann (1810–1882) proposed the **cell theory** in 1839. According to this theory, all life is composed of cells, cells arise only from preexisting cells, and the cell is the fundamental unit of structure and function in living organisms. Biologists interested in heredity began to examine cells to see what takes place in the course of cell reproduction. Walther Flemming (1843–1905) observed the division of chromosomes in 1879 and published a superb description of mitosis. By 1885, biologists generally recognized that the cell nucleus contains the hereditary information.

cell theory
Theory stating that all life is composed of cells, that cells arise only from other cells, and that the cell is the fundamental unit of structure and function in living organisms.

Charles Darwin (1809–1882), one of the most influential biologists of the nineteenth century, put forth the theory of evolution through natural selection and published his ideas in M rf c Mpgegj mdQncagccqin 1859. Darwin recognized that heredity was fundamental to evolution, and he conducted extensive genetic crosses with pigeons and other organisms. He never understood the nature of inheritance, however, and this lack of understanding was a major omission in his theory of evolution.

In the last half of the nineteenth century, cytologists demonstrated that the nucleus had a role in fertilization. Near the close of the nineteenth century, August Weismann (1834–1914) finally laid to rest the notion of the inheritance of acquired characteristics. He cut off the tails of mice for 22 consecutive generations and showed that the tail length in descendants remained stubbornly long. Weismann proposed the **germ-plasm theory**, which holds that the cells in the reproductive organs carry a complete set of genetic information that is passed to the egg and sperm (**Figure 1.10b**). This theory, and some of the other early theories of heredity that we have discussed up to this point, are summarized in **Table 1.1.**

germ-plasm theory
Theory stating that cells in the reproductive organs carry a complete set of genetic information.

TABLE 1.1 **Early concepts of heredity**

Concept	Proposed	Correct or incorrect
Pangenesis	Genetic information travels from different parts of the body to reproductive organs.	Incorrect
Inheritance of acquired	Acquired traits become incorporated into hereditary	Incorrect

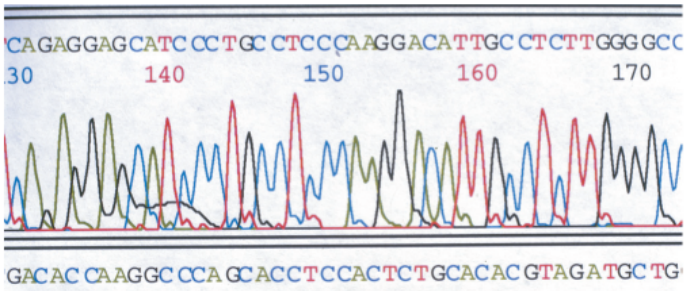
characteristics	information.	
Preformationism	Miniature organism resides in sex cells; thus all traits are inherited from one parent.	Incorrect
Blending inheritance	Genes blend and mix.	Incorrect
Germ-plasm theory	All cells contain a complete set of genetic information.	Correct
Cell theory	All life is composed of and cells arise only from cells.	Correct
Mendelian inheritance	Traits are inherited according to specific principles proposed by Mendel.	Correct

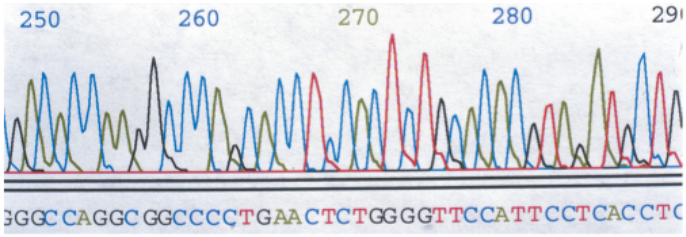
The year 1900 was a watershed in the history of genetics. Gregor Mendel's pivotal 1866 publication on his experiments with pea plants (discussed in more detail in Chapter 3), which revealed the principles of heredity, was rediscovered. Once the significance of his conclusions was recognized, other biologists immediately began to conduct similar genetic studies on mice, chickens, and other organisms. The results of these investigations showed that many traits indeed follow Mendel's rules.

In 1902, after the acceptance of Mendel's theory of heredity, Walter Sutton (1877–1916) proposed that genes are located on chromosomes. Thomas Hunt Morgan (1866–1945) discovered the first genetic mutant of fruit flies in 1910 and used fruit flies to unravel many details of transmission genetics. The foundation for population genetics was laid in the 1930s, when geneticists begin to integrate Mendelian genetics and evolutionary theory.

Geneticists began to use bacteria and viruses in the 1940s; the rapid reproduction and simple genetic systems of these organisms allowed detailed study of the organization and structure of genes. At about the same time, evidence accumulated that DNA was the repository of genetic information. James Watson (b. 1928) and Francis Crick (1916–2004), along with Maurice Wilkins (1916–2004) and Rosalind Franklin (1920–1958), described the three-dimensional structure of DNA in 1953, ushering in the era of molecular genetics (see Chapter 8).

By 1966, the chemical structure of DNA and the system by which it determines the amino acid sequence of proteins had been worked out. Advances in molecular genetics led to the first recombinant DNA experiments in 1973, which touched off another revolution in genetic research. Methods for rapidly sequencing DNA were first developed in 1977, which later allowed whole genomes of humans and other organisms to be determined. The polymerase chain reaction, a technique for quickly amplifying tiny amounts of DNA, was developed by Kary Mullis (b. 1944) and others in 1983. This technique is now the basis of numerous types of molecular analysis. In 1990, the Human Genome Project was launched. By 1995, the first complete DNA sequence of a free-living organism—the bacterium F_ck nmfgjsqdgęscd x_c—was determined, and the first complete sequence of a eukaryotic organism (yeast) was reported a year later. A rough draft of the human genome sequence was reported in 2000 (see Chapter 15), and the sequence was essentially completed in 2003, ushering in a new era in genetics (**Figure 1.13**). Today, the genomes of numerous organisms are being sequenced, analyzed, and compared. ▶ **TRY PROBLEMS 19 AND 20**





1.13 The human genome was completely sequenced in 2003. A chromatograph of a small part of the human genome.

The Future of Genetics

Numerous advances in genetics are being made today, and genetics remains at the forefront of biological research. New, rapid methods for sequencing DNA are being used to sequence the genomes of numerous species, from strawberries to butterflies to elephants. Recently, these methods were used to reconstruct the entire genome of an unborn fetus from fetal DNA circulating in the mother's blood, providing the potential for noninvasive prenatal genetic testing. Analysis of DNA from ancient bones demonstrates that several different species of humans roamed Earth as recently as 30,000 years ago. Powerful modern genetic techniques have been used to identify genes that influence agriculturally important characteristics, such as size in cattle, domestication in chickens, speed in racehorses, and leaf shape in corn. DNA analysis is now routinely used to identify and convict criminals or prove the innocence of suspects.

The power of new methods to identify and analyze genes is illustrated by genetic studies of myocardial infarction (heart attack) in humans. Physicians have long recognized that heart attacks run in families, but finding specific genes that contribute to an increased risk of heart attack has, until recently, been difficult. An international team of geneticists examined the DNA of 26,000 people in 10 countries for single nucleotide differences in their DNA (called single-nucleotide polymorphisms, or SNPs) that might be associated with an increased risk of heart attack. This and other similar studies identified several new genes that affect the risk of coronary artery disease and early heart attacks. These findings may make it possible to identify people who are predisposed to heart attack, allowing early intervention that might prevent the attacks. Analyses of SNPs are helping to locate genes that affect all types of traits, from eye color and height to glaucoma and cancer.

Information about sequence differences among organisms is also a source of new insights about evolution. For example, scientists analyzed DNA sequences at 26 genes to construct a comprehensive evolutionary tree of mammals. The tree revealed many interesting features of mammalian evolution; for instance, marine mammals (whales, dolphins, and porpoises) are most closely related to hippos.

In recent years, scientists have discovered that alterations to DNA and chromosome structure that do not involve the base sequence of the DNA play an important role in gene expression. These alterations, called epigenetic changes, affect our appearance, behavior, and health, and they are currently the focus of intense research. Other studies demonstrate that RNA is a key player in many aspects of gene function. The discovery in the late 1990s of tiny RNA molecules called small interfering RNAs and microRNAs led to the recognition that these molecules play central roles in gene expression and development. New genetic microchips that simultaneously analyze thousands of RNA molecules are providing information about the activities of thousands of genes in a given cell, allowing a detailed picture of how cells respond to external signals, environmental stresses, and diseases such as cancer. In the field of proteomics, powerful computer programs are being used to model the structure and function of proteins from DNA-sequence information. All this information provides us with a better understanding of numerous biological processes and evolutionary relationships. The flood of new genetic information, which requires the continuous

development of sophisticated computer programs to store, retrieve, compare, and analyze genetic data, has given rise to the field of bioinformatics, a merging of molecular biology and computer science.

As the cost of sequencing decreases, the focus of DNA-sequencing efforts will shift from the genomes of different species to individual differences within species. In the not-too-distant future, each person will probably possess a copy of his or her entire genome sequence, which can be used to help assess the risk of acquiring various diseases and to tailor their treatment should they arise. The use of genetics in agriculture will continue to improve the productivity of domestic crops and animals, helping to feed the future world population. This ever-widening scope of genetics raises significant ethical, social, and economic issues.

This brief overview of the history of genetics is not intended to be comprehensive; rather, it is designed to provide a sense of the accelerating pace of advances in genetics. In the chapters to come, we will learn more about the experiments and the scientists who helped shape the discipline of genetics.

THINK-PAIR-SHARE Question 5

CONCEPTS

Humans first applied genetics to the domestication of plants and animals between 10,000 and 12,000 years ago. Developments in plant hybridization and cytology in the eighteenth and nineteenth centuries laid the foundation for the field of genetics today. After Mendel's work was rediscovered in 1900, the science of genetics developed rapidly and today is one of the most active areas of science.

CONCEPT CHECK 3

How did developments in cytology in the nineteenth century contribute to our modern understanding of genetics?

1.3 A Few Fundamental Concepts Are Important for the Start of Our Journey into Genetics

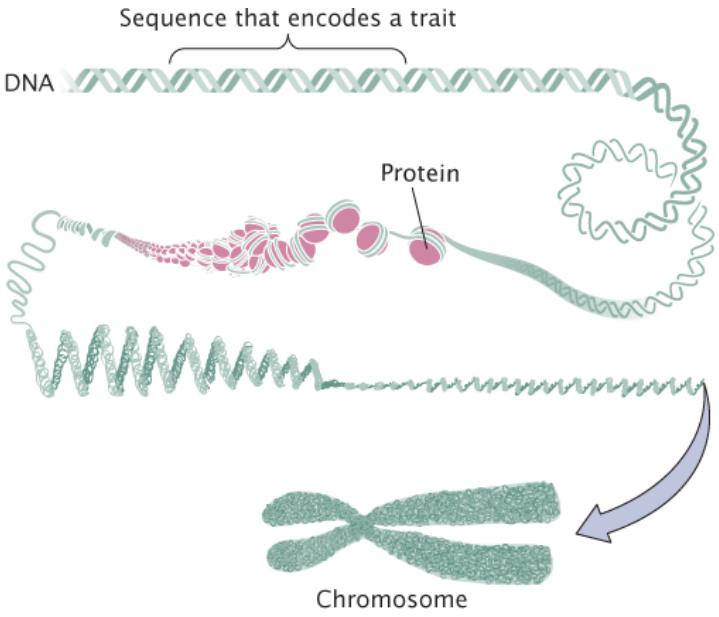
Undoubtedly, you learned some genetic principles in other biology classes. Let's take a few moments to review some fundamental genetic concepts.

- **Cells are of two basic types: eukaryotic and prokaryotic.** Structurally, cells consist of two basic types, although, evolutionarily, the story is more complex (see Chapter 2). Prokaryotic cells lack a nuclear membrane and do not generally possess membrane-bounded cell organelles, whereas eukaryotic cells are more complex, possessing a nucleus and membrane-bounded organelles such as chloroplasts and mitochondria.
- **The gene is the fundamental unit of heredity.** The precise way in which a gene is defined often varies depending on the biological context. At the simplest level, we can think of a gene as a unit of information that encodes a genetic characteristic. We will expand this definition as we learn more about what genes are and how they function.
- **Genes come in multiple forms called alleles.** A gene that specifies a characteristic may exist in several forms, called alleles. For example, a gene for coat color in cats may exist as an allele that encodes black fur or as an allele that encodes orange fur.
- **Genes confer phenotypes.** One of the most important concepts in genetics is the distinction between traits and genes. Traits are not inherited directly. Rather, genes are inherited and, along with environmental factors, determine the expression of traits. The genetic information that an individual organism possesses is its genotype; the trait is its phenotype. For example, the albinism seen in some Hopis is a phenotype, and the information in *OCA2* genes that causes albinism is the genotype.
- **Genetic information is carried in DNA and RNA.** Genetic information is encoded in the molecular structure of nucleic acids, which come in two types: deoxyribonucleic acid (DNA) and ribonucleic acid (RNA). Nucleic acids are polymers consisting of repeating units called nucleotides; each nucleotide consists of a sugar, a phosphate group, and a nitrogenous base. The nitrogenous bases in DNA are of four types: adenine (A), cytosine (C), guanine (G), and thymine (T). The sequence of these bases encodes genetic information. DNA consists of two complementary nucleotide strands. Most organisms carry their genetic information in DNA, but a few viruses carry it in RNA. The four nitrogenous bases of RNA are adenine, cytosine, guanine, and uracil (U).

THINK-PAIR-SHARE Question 6

- **Genes are located on chromosomes.** The vehicles of genetic information within a cell are chromosomes (**Figure 1.14**), which consist of DNA and associated proteins. The cells of each species have a characteristic number of chromosomes; for example, bacterial cells normally possess a single chromosome; human cells possess 46; pigeon cells possess 80. Each chromosome carries a large number of genes.
- **Replicated chromosomes separate through the processes of mitosis and meiosis.** The processes of mitosis and meiosis ensure that a complete set of an organism's chromosomes exists in each cell resulting from cell division. Mitosis is the separation of replicated chromosomes in the division of somatic (nonsex) cells. Meiosis is the pairing and separation of replicated chromosomes in the division of sex cells to produce gametes (reproductive cells).

- **Genetic information is transferred from DNA to RNA to protein.** Many genes encode characteristics by specifying the structure of proteins. Genetic information is first transcribed from DNA into RNA, and then RNA is translated into the amino acid sequence of a protein.
- **Mutations are changes in genetic information that can be passed from cell to cell or from parent to offspring.** Gene mutations affect the genetic information of only a single gene; chromosome mutations alter the number or the structure of chromosomes and therefore usually affect many genes.
- **Many traits are affected by multiple factors.** Many traits are affected by multiple genes that interact in complex ways with environmental factors. Human height, for example, is affected by many genes as well as by environmental factors such as nutrition.
- **Evolution is genetic change.** Evolution can be viewed as a two-step process: first, genetic variation arises, and second, some genetic variants increase in frequency, whereas other variants decrease in frequency. **TRY PROBLEM 21**



Pierce, *Genetics Essentials: Concepts and Connections*, 4e, © 2018
W. H. Freeman and Company

1.14 Genes are carried on chromosomes.

COMPREHENSION QUESTIONS

Answers to questions and problems preceded by an asterisk can be found at the end of the book.

Section 1.1

*1. How did Hopi culture contribute to the high incidence of albinism among members of the Hopi tribe?

2. Give at least three examples of the role of genetics in society today.

3. Briefly explain why genetics is crucial to modern biology.

4. List the three traditional subdisciplines of genetics and summarize what each covers.

5. What are some characteristics of model genetic organisms that make them useful for genetic studies?

Section 1.2

6. When and where did agriculture first arise? What role did genetics play in the development of the first domesticated plants and animals?

7. Outline the concept of pangenesis and explain how it differs from the germ-plasm theory.

8. What does the concept of the inheritance of acquired characteristics propose and how is it related to the notion of pangenesis?

9. What is preformationism? What did it have to say about how traits are inherited?

10. Define blending inheritance and contrast it with preformationism.

11. How did developments in botany in the seventeenth and eighteenth centuries contribute to the rise of modern genetics?

12. List some advances in genetics made in the twentieth century.

13. Briefly explain the contribution that each of the following people made to the study of genetics.

a. **Matthias Schleiden and Theodor Schwann**

b. **August Weismann**

c. **Gregor Mendel**

d. **James Watson and Francis Crick**

e. **Kary Mullis**

Section 1.3

14. What are the two basic cell types (from a structural perspective) and how do they differ?

15. Summarize the relations between genes, DNA, and chromosomes.

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

APPLICATION QUESTIONS AND PROBLEMS

Section 1.1

*16. How are genetics and evolution related?

*17. For each of the following genetic topics, indicate whether it focuses on transmission genetics, molecular genetics, or population genetics.

a. **Analysis of pedigrees to determine the probability of someone inheriting a trait.**

b. **Study of people on a small island to determine why a genetic form of asthma is so prevalent on the island.**

c. **Effect of nonrandom mating on the distribution of genotypes among a group of animals.**

d. **Examination of the nucleotide sequences found at the ends of chromosomes.**

e. **Mechanisms that ensure a high degree of accuracy during DNA replication.**

f. **Study of how the inheritance of traits encoded by genes on sex chromosomes (sex-linked traits) differs from the inheritance of traits encoded by genes on nonsex chromosomes (autosomal traits).**

Section 1.2

*18. Genetics is said to be both a very old science and a very young science. Explain what this means.

*19. Match each description (_ through b) with the correct theory or concept listed below.

a. **Each reproductive cell contains a complete set of genetic information.**

b. **All traits are inherited from one parent.**

c. **Genetic information may be altered by the use of a characteristic.**

d. **Cells of different tissues contain different genetic information.**

Preformationism
Pangenesis
Germ-plasm theory
Inheritance of acquired characteristics

*20. Compare and contrast the following ideas about inheritance.

a. **Pangenesis and germ-plasm theory.**

b. **Preformationism and blending inheritance.**

c. **The inheritance of acquired characteristics and our modern theory of heredity.**

Section 1.3

*21. Compare and contrast the following terms:

a. **Eukaryotic and prokaryotic cells**

b. **Gene and allele**

c. **Genotype and phenotype**

d. **DNA and RNA**

e. **DNA and chromosome**

CHALLENGE QUESTIONS

Introduction

*22. The type of albinism that arises with high frequency among the Hopis (discussed in the introduction to this chapter) is most likely oculocutaneous albinism type 2, which is caused by a defect in the MA? 0 gene on chromosome 15. Do some research on the Internet to determine how the phenotype of this type of albinism differs from the phenotypes of other

forms of albinism in humans and the mutated genes that result in those phenotypes. Hint: Visit the website Online Mendelian Inheritance in Man and search the database for albinism.

Section 1.1

23. We now know a great deal about the genetics of humans. What are some of the reasons humans have been the focus of intensive genetic study?

24. Describe some of the ways in which your own genetic makeup affects you as a person. Be as specific as you can.

25. Describe at least one trait that appears to run in your family (appears in multiple members of the family). Do you think that this trait runs in your family because it is an inherited trait or because it is caused by environmental factors that are common to family members? How might you distinguish between these possibilities?

Section 1.3

***26.** Suppose that life exists elsewhere in the universe. All life must contain some type of genetic information, but alien genomes might not consist of nucleic acids and have the same features as those found in the genomes of life on Earth. What do you think might be the common features of all genomes, no matter where they exist?

27. Choose one of the ethical or social issues in _ through b below and give your opinion on the issue. For background information, you might read one of the articles on ethics listed and marked with an asterisk in the Suggested Readings for Chapter 1 in your SaplingPlus.

a. Should a person's genetic makeup be used in determining his or her eligibility for life insurance?

b. Should biotechnology companies be able to patent newly sequenced genes?

c. Should gene therapy be used on people?

d. Should genetic testing for inherited disorders for which there is no treatment or cure be made available?

***28.** A 45-year-old woman undergoes genetic testing and discovers that she is at high risk for developing colon cancer and Alzheimer disease. Because her children have 50% of her genes, they may also be at an increased risk for these diseases. Does she have a moral or legal obligation to tell her children and other close relatives about the results of her genetic testing?

***29.** Suppose that you could undergo genetic testing at age 18 for susceptibility to a genetic disease that would not appear until middle age and has no available treatment.

a. What would be some of the possible reasons for having or not having such a genetic test?

b. Would you personally want to be tested? Explain your reasoning.

THINK-PAIR-SHARE QUESTIONS

Think-Pair-Share questions are designed to be worked in collaboration with other students. First THINK about the question, then PAIR up with one or more other students, and finally SHARE your answers and work together to arrive at a solution.

Introduction

1. Albinism occupied a special place in the Hopi culture; individuals who possessed this trait were valued by members of the tribe. What are some examples of genetic traits that, in contrast, sometimes result in discrimination and prejudice?

2. **Albinism in humans can be caused by mutations in any one of several different genes. This situation, in which the same phenotype may result from variation in several different genes, is referred to as genetic heterogeneity. Is genetic heterogeneity common? Are most genetic traits in humans the result of variation in a single gene, or are there many genetic traits that result from variation in several genes, as albinism does?**

Section 1.1

3. **Bob says that he is healthy and has no genetic diseases such as hemophilia or Down syndrome. Therefore, he says, genetics plays little role in his life. Do you think Bob is correct in his conclusion? Why or why not?**
4. **Are mutations good or bad? Explain your answer.**

Section 1.2

5. **Do you support or oppose the development of genetically engineered foods (genetically modified organisms, or GMOs)? Find someone who takes the opposite position, and discuss this question with him or her. Think about the economic and environmental benefits, health risks, ecological effects, and social impact of the use of genetically engineered foods. List some reasons for and against genetically engineering the foods we eat.**

Section 1.3

6. **Why do you think all organisms use nucleic acids for encoding genetic information? Why not use proteins or carbohydrates? What advantages might DNA have as the source of genetic information?**

CONCEPTS SUMMARY

- Genetics is central to the life of every person: it influences a person's physical features, personality, intelligence, and susceptibility to numerous diseases.
- Genetics plays important roles in agriculture, the pharmaceutical industry, and medicine. It is key to the study of biology.
- All organisms use similar genetic systems. Genetic variation is the foundation of evolution and is critical to understanding all life.
- The study of genetics can be divided into transmission genetics, molecular genetics, and population genetics.
- Model genetic organisms are species about which much genetic information exists because of characteristics that make them particularly amenable to genetic analysis.
- The use of genetics by humans began with the domestication of plants and animals.
- The ancient Greeks developed the concepts of pangenesis and the inheritance of acquired characteristics, both of which were later disproved.
- Preformationism suggested that a person inherits all of his or her traits from one parent. Blending inheritance proposed that offspring possess a mixture of the parental traits. These ideas were later shown to be incorrect.
- By studying the offspring of crosses between varieties of peas, Gregor Mendel discovered the principles of heredity. Developments in cytology in the nineteenth century led to the understanding that the cell nucleus is the site of heredity.
- In 1900, Mendel's principles of heredity were rediscovered. Population genetics was established in the early 1930s, followed closely by biochemical genetics and bacterial and viral genetics. The structure of DNA was discovered in 1953, stimulating the rise of molecular genetics.
- There are two basic types of cells: prokaryotic and eukaryotic.
- The genes that determine a trait are termed the genotype; the trait that they produce is the phenotype.
- Genes are located on chromosomes, which are made up of nucleic acids and proteins and are partitioned into daughter cells through the process of mitosis or meiosis.
- Genetic information is expressed through the transfer of information from DNA to RNA to proteins.
- Evolution requires genetic change in populations.

IMPORTANT TERMS

genome (p. 4)
transmission genetics (p. 5)
molecular genetics (p. 5)
population genetics (p. 5)
model genetic organism (p. 5)

ANSWERS TO CONCEPT CHECKS

1. **d**
2. **No, because horses are expensive to house, feed, and propagate, they have too few progeny, and their generation time is too long.**
3. **Developments in cytology in the 1800s led to the identification of parts of the cell, including the cell nucleus and chromosomes. The cell theory focused biologists' attention on the cell, eventually leading to the conclusion that the nucleus contains the hereditary information.**