CHAPTER 11

Human Heredity





In humans, as in all living things, traits such as sex and skin color are inherited.

Of all the organisms in the world, there is one that we find particularly fascinating. This organism is not, however, an ideal one for the study of genetics. Why? The reasons are many. It is composed of approximately 10^{14} (100 million million) cells. Its generation time (about 20 years) is much too long. It cannot be kept in the laboratory for study. It produces too few offspring for good statistics. It cannot be used in test crosses. And its genetic system contains nearly 100,000 genes. Despite these disadvantages, biologists have studied the genetics of this single organism, Homo sapiens, for more than 80 years. The reason for so much scientific interest is clear: Of all the organisms on Earth, it is ourselves we long to know the best. In this chapter, you will discover how some human traits are inherited and how some inherited disorders can be cured or prevented.

CHAPTER OUTLINE

11–1 "It Runs in the Family"
- The Human Organism
- Human Traits

11–2 The Inheritance of Human Traits
- Human Blood Groups
- Huntington Disease
- Sickle Cell Anemia
- Polygenic Traits

11–3 Sex-Linked Inheritance
- Sex Determination
- Sex-Linked Genetic Disorders
- Sex-Influenced Traits

11–4 Diagnosis of Genetic Disorders
- A Chromosomal Abnormality—Down Syndrome
- Prenatal Diagnosis
- Ethical Considerations

11–1 "It Runs in the Family"

Section Objectives

- Explain how human traits are inherited.
- Distinguish between sex chromosomes and autosomes.
- Discuss the influence of the environment on gene expression.

How many times have you overheard two people discussing whether a newborn baby looks more like its mother or its father? People have always discussed such things. But when those of us who study biology engage in such discussions, we make an important assumption: Human children inherit characteristics from their parents as a result of gene interactions.

The principles of genetics that we have already discussed apply to humans. **Many human traits are inherited by the action of dominant and recessive genes, although other traits are determined through more complicated gene interactions.** Although the study of human genetics is difficult, it is one of the most important fields in biology. For we are interested in finding out as much as possible about ourselves, including the role that heredity plays in determining who and what we are and how some of the disorders and conditions that we inherit can be cured or prevented.

The Human Organism

The study of ourselves logically begins with a discussion of human chromosomes. Recall that a diploid cell has two sets of homologous chromosomes. A human diploid cell contains 46 chromosomes arranged in 23 pairs. These 46 chromosomes

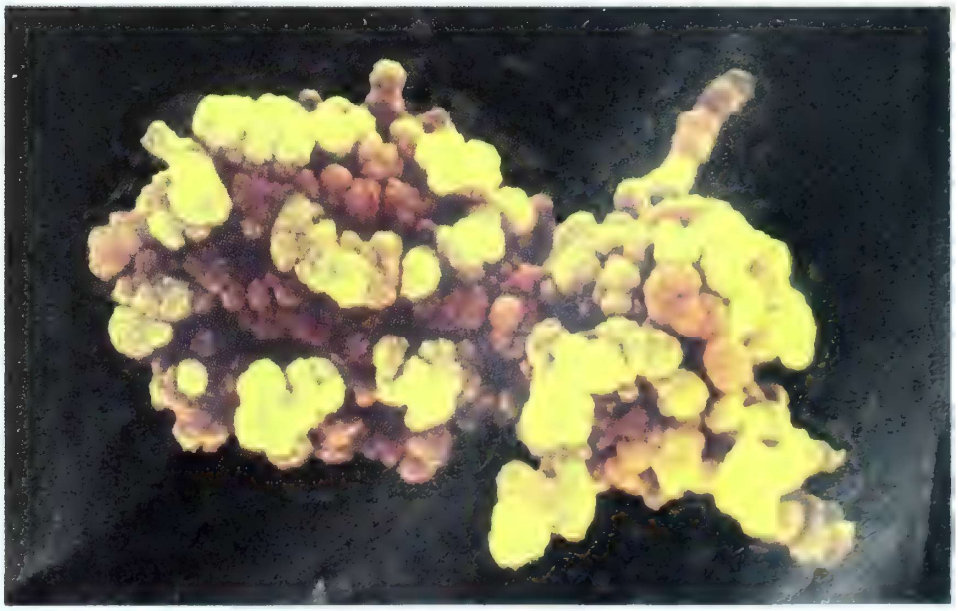


Figure 11–1 *Each human chromosome contains a specific set of genetic instructions that is passed on from parent to child. The human chromosome in this electron micrograph has been magnified approximately 20,000 times.*

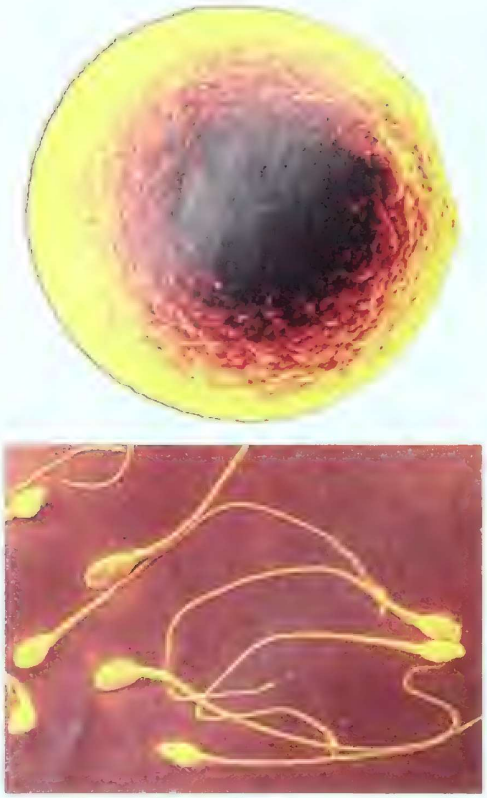


Figure 11–2 *In humans, the egg (top) and the sperm (bottom) contain 23 chromosomes each. During fertilization, these gametes unite to produce a zygote that contains 46 chromosomes.*

contain 6 billion nucleotide pairs of DNA—6 billion individual characters of the genetic code. To get an idea of how long a complete human DNA sequence actually is, consider the following: In this textbook, there are approximately 3000 letters on each page. If a complete human DNA sequence were to be written in the same-size type as this textbook, it would comprise a book more than 1 million pages long!

As you will recall from Chapter 9, the principles of genetics described by Mendel require that organisms inherit a single copy of each gene from each parent. In humans, the **gametes**, or reproductive cells, contain a single copy of each gene. Gametes (sperm and eggs) are formed in the reproductive organs by the process of meiosis. Each egg cell and each sperm cell contain 23 chromosomes, or the haploid number of chromosomes. During fertilization, sperm and egg unite and a **zygote** (ZIGH-goht), or fertilized egg, is produced. The zygote contains 46 chromosomes (23 pairs), or the diploid number of chromosomes characteristic of the organism.

Of the 46 chromosomes found in a human diploid cell, two are the sex chromosomes, X and Y. The remaining 44 chromosomes are the autosomes. The inheritance of dominant and recessive human genes carried on any of the 44 autosomes follows Mendel's principles. Human genes carried on the sex chromosomes are sex-linked.

The simplest patterns of inheritance occur in traits that are influenced by a single gene. Fortunately, many such genes have now been described. Human genetics is advancing so rapidly that scarcely a week goes by without a new human gene being reported and mapped.

Human Traits

You will recall from Chapter 10 that the phenotype of an organism is only partly determined by its genotype. There are some traits that are strongly influenced by environmental, or nongenetic, factors. Environmental factors include nutrition and exercise. For example, recent studies show that improvements in infant and childhood nutrition in the twentieth century in the United States and Europe have greatly increased



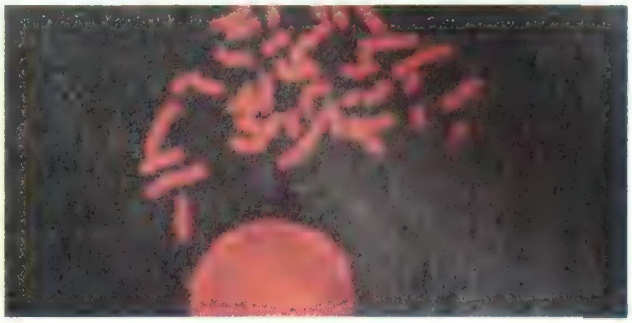
Figure 11–3 *Notice the difference in the heights of these teenagers. Although the genes for height are inherited, the trait is strongly influenced by environmental factors, such as diet.*

SCIENCE, TECHNOLOGY, AND SOCIETY **BREAKTHROUGH**

How Do We Map Human Genes?

Preparing a human genetic map presents special challenges. As we saw in Chapter 10, crossing-over is the key to gene mapping in most organisms: Crossovers rarely occur between nearby genes but are common between distant genes. It is possible to map human genes in this way, but the need to gather crossover data from several generations has made human gene mapping slow and tedious. In the last few years, however, several breakthroughs have made it possible to map human genes much more rapidly.

One of the most exciting new techniques was developed by Jeanne Lawrence and Robert Singer at the University of Massachusetts Medical School. Their technique attaches a fluorescent label to pieces of DNA complementary to a particular gene. Once the labeled DNA fragments are mixed with a chromosome preparation, they attach to the DNA of the gene in question by a chemical bond known as a hydrogen bond. This action produces an intense fluorescent label at the place on the chromosome where the gene is located. The Lawrence-Singer technique is just one more tool in an increasingly powerful workshop for exploring the human genetic system.



Two tiny yellow spots mark the location of the genes of the Epstein-Barr virus.

the average height of these populations over the nineteenth-century averages. Famine-stricken countries, however, were found to produce a generation with stunted growth.

Although it is important to consider the influence of the environment on the expression of some genes, it must be understood that environmental effects on gene expression are not inherited; genes are. Genes that are denied a proper environment in which to reach full expression in one generation can, in a proper environment, achieve full potential in a later generation.

11-1 SECTION REVIEW

1. How are human traits inherited?
2. What is a gamete? A zygote?
3. How many chromosomes are in a human diploid cell? A human haploid cell?
4. Distinguish between sex chromosomes and autosomes.
5. How does the environment influence gene expression?

Section Objectives

- Identify two human traits that are determined by multiple alleles.
- Explain how Huntington disease and sickle cell anemia are inherited.
- Distinguish between traits determined by multiple alleles and polygenic traits.

11–2 The Inheritance of Human Traits

At the writing of this textbook, more than 1000 human genes have been described and studied. Listing all of these genes would serve no purpose, but it is useful and interesting to examine a few cases in which particular genes affect us in very significant, and often dramatic, ways.

Human Blood Groups

Multiple alleles are three or more alleles of the same gene that code for a single trait. ABO and Rh blood groups are determined by multiple alleles. A number of other human traits are determined by **multiple alleles.**

ABO BLOOD GROUPS In 1900, the Austrian physician Karl Landsteiner discovered that human blood could be classified into four general groups, or types, known today as the Landsteiner blood groups. These four blood types are determined by the presence or absence of specific chemical substances in the blood.

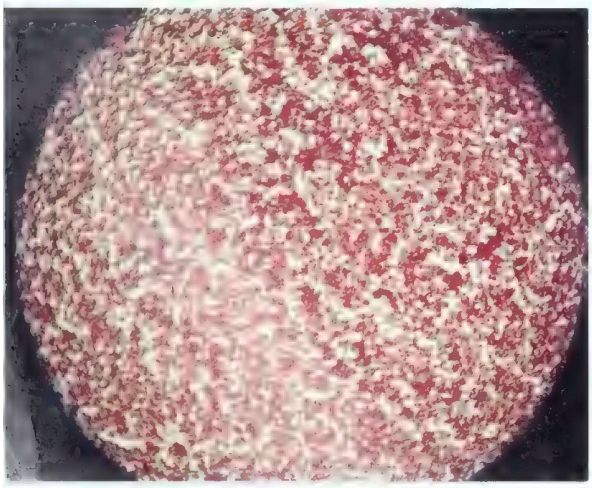
Landsteiner observed that when he mixed together the red blood cells from different people, the red blood cells agglutinated, or clumped together. After doing extensive research, Landsteiner determined that the red blood cells contained two antigens, A and B. Antigens are molecules that can be recognized by the immune system.

The presence or absence of these antigens (A and B) produces four possible blood types: A, B, AB, and O. A person with type A blood has red blood cells that contain antigen A but not antigen B. A person with type B blood has red blood cells that contain antigen B but not antigen A. A person with type AB blood has both antigen A and antigen B. And a person with type O blood has neither antigen A nor antigen B.

The ABO blood groups are of particular importance in blood transfusions. A transfusion of the wrong blood type can cause a violent, often fatal, reaction in the body as the immune system responds to the presence of an antigen not found on its own cells. For example, if type B blood is given to a person with type A or type O blood, a reaction will occur against the red blood cells containing the B antigen. Because people with type AB blood have both antigens on their red blood cells, they can receive blood from any of the four types.

The Landsteiner, or ABO, blood groups are determined by a single gene with three possible alleles: I^A, I^B, or i. Figure 11–5 illustrates the relationship of these alleles to the blood group phenotype. Because the I^A and I^B alleles are both expressed when they occur together, they are said to be codominant.

Figure 11–4 This photomicrograph shows the agglutination, or clumping, of human red blood cells that can occur when blood of two different types is mixed.



Rh BLOOD GROUPS In addition to the ABO antigens, there is another antigen on the red blood cells that determines the success or failure of transfusions. It is the Rh antigen (named after the rhesus monkey in which the antigen was first discovered by Landsteiner and Alexander Wiener). People who have the Rh antigen on their red blood cells are said to be Rh positive (Rh^+). People who do not have the Rh antigen on their red blood cells are said to be Rh negative (Rh^-). It is interesting to note that although the Rh blood groups are commonly referred to as either positive or negative, there are actually more than two alleles that determine these blood groups. In fact, there are eight common alleles and many that are rarer.

In blood banks, the ABO and Rh blood groups are often expressed together in symbols such as AB^- or O^+. The gene for the Rh antigen, however, displays simple dominance. Genotype Rh^+ Rh^+ and Rh^- Rh^+ produces the Rh positive phenotype. Only the double recessive genotype Rh^- Rh^- produces Rh negative blood.

Genotypes	Phenotypes
ii	O
$I^A I^A$, $I^A i$	A
$I^B I^B$, $I^B i$	B
$I^A I^B$	AB

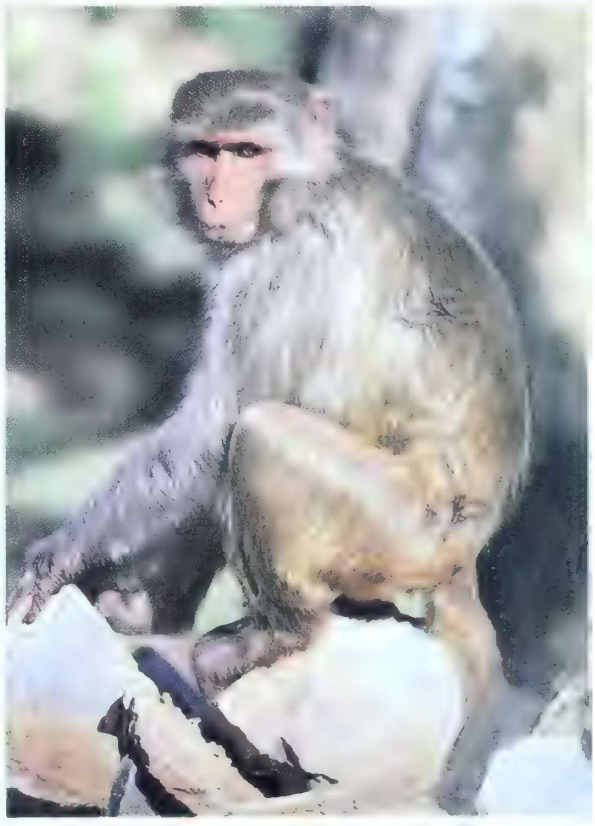
Figure 11–5 *This chart illustrates the relationship between genotype and phenotype for each of the ABO blood groups. What are the possible genotypes of a person with phenotype B blood?*

Huntington Disease

Many of the tens of thousands of human genes are so essential to the normal functioning of cells and tissues that we do not notice them unless something goes wrong and a genetic disease results. **Huntington disease, which is produced by a single dominant allele (H), is an example of a genetic disease.** People who have this disease show no symptoms until they are in their thirties or forties, when the gradual damage to their nervous system begins. As you will recall from Chapter 10, the effects of a dominant allele are expressed even when the recessive allele is present. So people who have the dominant allele for Huntington disease have the disease and suffer painful progressive loss of muscle control and mental function until death occurs.

Until very recently, the only way people knew if they carried the gene for Huntington disease was the appearance of the disease later in their lives. Thus many Huntington disease sufferers had children who inherited the gene for this disease.

Figure 11–6 *This photograph shows a rhesus monkey, the type of monkey after which the Rh blood group was named.*



Sickle Cell Anemia

In 1904, Doctor James Herrick noticed an unusual ailment afflicting one of his young patients. His patient, a 20-year-old black college student, had been complaining of weakness and dizzy spells and had open sores on his legs. The Chicago physician examined the student's blood cells under a microscope and discovered that many of the red blood cells were bent and twisted into shapes that resembled sickles (farm tools used to cut grain). The normal shape of a red blood cell is that of a round, flattened disk. Doctor Herrick guessed correctly that these unusually shaped cells were the cause of his patient's



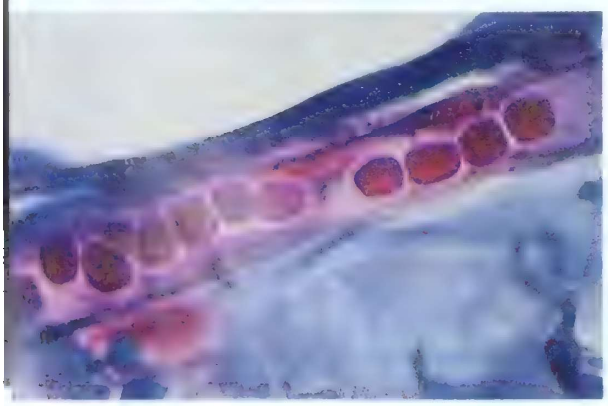
Figure 11–7 *These scanning electron micrographs show human red blood cells that contain normal hemoglobin (left) and a red blood cell that contains the abnormal hemoglobin characteristic of sickle cell anemia (right). The red blood cells have been magnified approximately 10,000 times.*

problems, and he gave the disease the name by which we know it today—sickle cell anemia.

THE CAUSE OF SICKLE CELL ANEMIA Sickle cell anemia is caused by a change in one of the polypeptides found in hemoglobin. Hemoglobin is the protein that carries oxygen in red blood cells.

When a person who has sickle cell anemia is deprived of oxygen (from heavy exercise, holding one's breath, or even nervousness or anxiety), the hemoglobin molecules join together and form fibers. These fibers cause the red blood cells to undergo the dramatic changes in shape that Doctor Herrick observed. The sickle-shaped red blood cells are more rigid and tend to become stuck in the capillaries, the narrowest blood vessels in the body. See Figure 11–8. As a result, the movement of blood through these vessels is stopped and damage to cells and tissues occurs. Serious injury or death may result.

Figure 11–8 *Capillaries, which are the narrowest blood vessels, are so small that they permit only one red blood cell at a time to pass through. What happens to sickle-shaped red blood cells as they move through the capillaries?*



THE GENETICS OF SICKLE CELL ANEMIA **The gene for normal hemoglobin (A) is codominant with the sickle cell gene (S).** People who are heterozygous (AS) are said to be sickle cell carriers. Because roughly half of the hemoglobin molecules in the blood of carriers is normal, these people suffer few ill effects of the disorder. People who are homozygous (SS) are said to be sickle cell sufferers. Because all of their hemoglobin molecules are affected by the sickle cell gene, these people are more severely afflicted by the disease.

THE MOLECULAR BASIS OF SICKLE CELL ANEMIA The gene for sickle cell hemoglobin differs from the gene for normal hemoglobin by a single nucleotide. Why does this seemingly small difference cause so much trouble?

The substitution of one nucleotide in the gene results in the substitution of a different amino acid in the sickle cell

hemoglobin protein. This change makes hemoglobin less soluble in water. Most of the time the condition does not present a problem. But when the body is under even minor stress, the hemoglobin in a large proportion of red blood cells will come out of solution as crystals. This crystallization of hemoglobin causes the sickle-shaped cells to appear, accompanied by their medical consequences.

THE DISTRIBUTION OF SICKLE CELL ANEMIA In the United States, people of African ancestry are the most common carriers of the sickle cell trait. In the rest of the world, sickle cell anemia is found in the tropical regions of Africa and Asia. Approximately 10 percent of Americans of African ancestry and as many as 40 percent of the population in some parts of Africa carry the trait. Why is sickle cell anemia so common in some regions and virtually unknown in others (such as Northern Europe and Asia)?

The answer to this question provides us with a surprising lesson in evolution. People who are heterozygous for sickle cell anemia (AS) are partially resistant to malaria, a serious disease that affects the red blood cells. The sickle cell hemoglobin offers this resistance because the malaria parasite, which lives within the red blood cell, can be killed when hemoglobin molecules crystallize out of solution. People who are homozygous for normal hemoglobin (AA), on the other hand, have no resistance to malaria.

The incidence of sickle cell anemia parallels the incidence of malaria throughout the tropical areas of the world. The sickle cell trait probably developed in many people throughout the world as a single mutation, or change in genetic material. That mutation conferred an advantage wherever malaria was common, and thus it was favored by natural selection. Sickle cell anemia has persisted wherever it has helped its carriers survive malaria.

	A	S
A	AA	AS
A	AA	AS

A = gene for normal hemoglobin
S = gene for sickle cell hemoglobin

Figure 11–9 *This Punnett square shows the cross between a person who is heterozygous for sickle cell anemia and a person who is homozygous for normal hemoglobin. What are the possible genotypes of their offspring?*



Figure 11–10 *Two of the red blood cells in this scanning electron micrograph have been invaded by the parasite that causes malaria. People who are heterozygous for sickle cell anemia are partially resistant to malaria.*



Figure 11–11 *The color of human skin ranges from very light to very dark, depending upon the amount of melanin (dark pigment) present in the skin cells. Very dark-skinned people have alleles that code for production of melanin at all the gene positions for skin color. In very light-skinned people fewer of these positions are occupied by alleles that code for melanin production.*

Polygenic Traits

Human traits that are controlled by a number of genes are called **polygenic** (pahl-uh-JEHN-ihk) **traits**. Examples of polygenic human traits include height, body weight, and skin color.

Unlike the simple skin pigmentation system of many animals, which allows a single gene to determine color, the human skin pigmentation system is rather complex. In humans, at least four different genes control skin color, and several of these genes may have multiple alleles. This means that the inheritance of skin color in our species can be somewhat unpredictable. And because skin color is a polygenic trait, children of the same mother and father may have quite different patterns of skin color—each pattern determined by which combination of genes they have inherited from their parents.

The color of human skin ranges from very dark to very light, depending upon the amount of melanin, a dark-colored pigment, present in skin cells. Very dark-skinned people have alleles that code for the production of melanin at all their gene positions for skin color. Light-skinned people, on the other hand, have alleles that code for the production of melanin at fewer gene positions for skin color. The range of human skin color, spread across the globe with no clear dividing line, is an illustration of what a wonderfully diverse species we are.

11-2 SECTION REVIEW

1. List several human traits determined by multiple alleles.
2. How is Huntington disease inherited?
3. What is sickle cell anemia?
4. What are polygenic traits? Give two examples of polygenic traits.
5. If a man with type O blood marries a woman with type AB blood, what are the possible genotypes of their offspring?

11–3 Sex-Linked Inheritance

Section Objectives

- Describe how sex is determined in humans.
- List two conditions of nondisjunction of sex chromosomes.
- Identify some human sex-linked traits.
- Compare sex-linked and sex-influenced traits.

As you will recall from Chapter 10, genes that are located on the sex chromosomes of an organism are inherited in a sex-linked pattern. As in many organisms, the sex in humans is determined by the X and Y chromosomes. In females, meiosis produces egg cells that contain one X chromosome and 22 autosomes. In males, meiosis produces sperm cells of which half contain one Y chromosome and 22 autosomes and the other half contain one X chromosome and 22 autosomes.

The sex of a person is determined by whether an egg cell is fertilized by an X-carrying sperm or a Y-carrying sperm. Males are normally 46XY, meaning that they have a total of 46 chromosomes, including an X chromosome and a Y chromosome. Females are normally 46XX.

Sex Determination

How do the X and Y chromosomes determine whether the sex of the zygote will be male or female? In *Drosophila* (fruit flies), which also have an XY system of sex determination, geneticists have found that sex is determined by the number of X chromosomes. A fruit fly that has a single X chromosome is male, regardless of whether it has a Y chromosome. A fruit fly that has two X chromosomes is female, regardless of whether a Y chromosome is present. This is not so in humans.

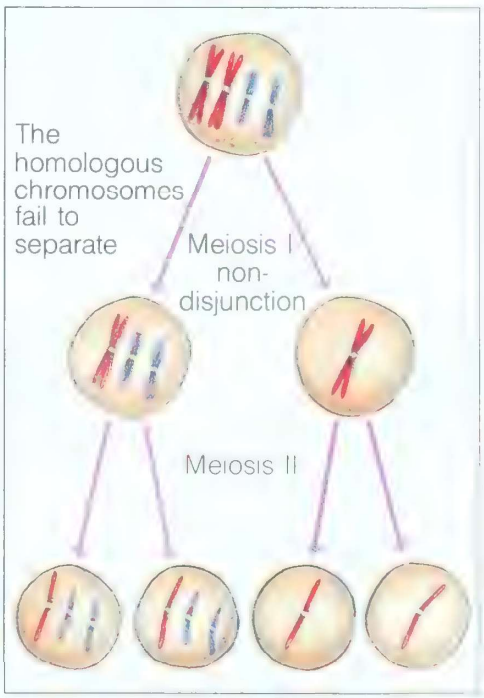
THE HUMAN XY SYSTEM Although meiosis is a precise mechanism that separates the two sex chromosomes of a diploid cell into single chromosomes of haploid gamete cells, errors sometimes do take place. The most common of these errors is nondisjunction. **Nondisjunction is the failure of chromosomes to separate properly during one of the stages of meiosis.** See Figure 11–12.

Nondisjunction can produce gametes that contain either two sex chromosomes or no sex chromosomes—a direct contrast to the normal condition of one sex chromosome. When one of these gametes joins with a normal gamete during fertilization, the result is a person with an abnormal number of sex chromosomes.

NONDISJUNCTION DISORDERS Roughly 1 birth in 1000 is affected by an abnormality involving nondisjunction of the sex chromosomes. The most common abnormalities are Turner syndrome and Klinefelter syndrome.

People who have Turner syndrome are female in appearance but their female sex organs do not develop at puberty and they are sterile, or unable to have children. Turner syndrome is abbreviated 45X or 45XO, where O denotes the absence of a second sex chromosome. People with Klinefelter syndrome are

Figure 11–12 *When homologous chromosomes fail to separate during meiosis, nondisjunction results.*



Zeroing in on the Maleness Gene

Disorders such as Turner syndrome and Klinefelter syndrome make it clear that maleness is determined by a gene on the Y chromosome. But how would one go about finding that gene? The crucial first steps have now been taken by David Page, a young researcher at the Massachusetts Institute of Technology (MIT). As he studied human sex determination, Page learned that doctors had been treating a number of male patients whose genotypes were 46XX. In every case, these people were found to have portions of the Y chromosome attached to one of their X chromosomes. Page reasoned that these portions must contain the gene for male determination.



Each human has traits that result from the expression of inherited genes. Today, scientists are studying the genetic makeup of X and Y chromosomes to learn how maleness and femaleness are determined.

By analyzing the Y-chromosome portions, Page and his associates found that a single identical piece of the Y chromosome, about 100,000 DNA bases in length, was present in every one of the XX males. Investigators in many laboratories are now concentrating on the structure of this 100,000-base region. Surprisingly, a section of this region is almost identical to a normal fragment found on the X chromosome. No one is certain why the X and Y chromosomes should have a nearly identical fragment, but every new piece of information brings us that much closer to understanding one of the most fundamental of all human differences—maleness and femaleness.

male in appearance, and they, too, are sterile. The abbreviation for Klinefelter syndrome is 47XXY.

It is interesting to note that there have been no reported instances of babies being born without an X chromosome. Embryos without an X chromosome do not seem to develop properly because, as we will soon discover, the X chromosome contains a number of genes that are vital for normal development.

What can we learn from these abnormalities of the sex chromosomes? First, an X chromosome is absolutely essential for survival. Second, sex seems to be determined by the presence or absence of a Y chromosome and not by the number of X chromosomes. For example, there have been reported cases of people who have genotypes 48XXXY and 49XXXXY and are male in appearance.

After years of uncertainty, the reason for the genetic importance of the Y chromosome is finally becoming clear: The Y chromosome contains a gene that switches on the male pattern of growth during embryological development. If this gene is absent, the embryo follows a female pattern of growth.

Sex-Linked Genetic Disorders

Genes that are carried on either the X or the Y chromosome are said to be **sex-linked**. In humans, the small Y chromosome carries very few genes. The much larger X chromosome contains a number of genes that are vital to proper growth and development. And as you just read, it seems to be impossible for humans to develop without the genes of the X chromosome.

It is particularly easy to spot recessive defects in genes located on the X chromosome because the genes are expressed more commonly in males than in females. What is the reason for this? Recall that males have one X chromosome. Thus all X-linked genes are expressed in males, even if they are recessive. In order for a recessive gene to be expressed in females, there must be two copies of it, one on each of the two X chromosomes. If one of the X chromosomes contains a dominant gene, it will mask the expression of the recessive gene.

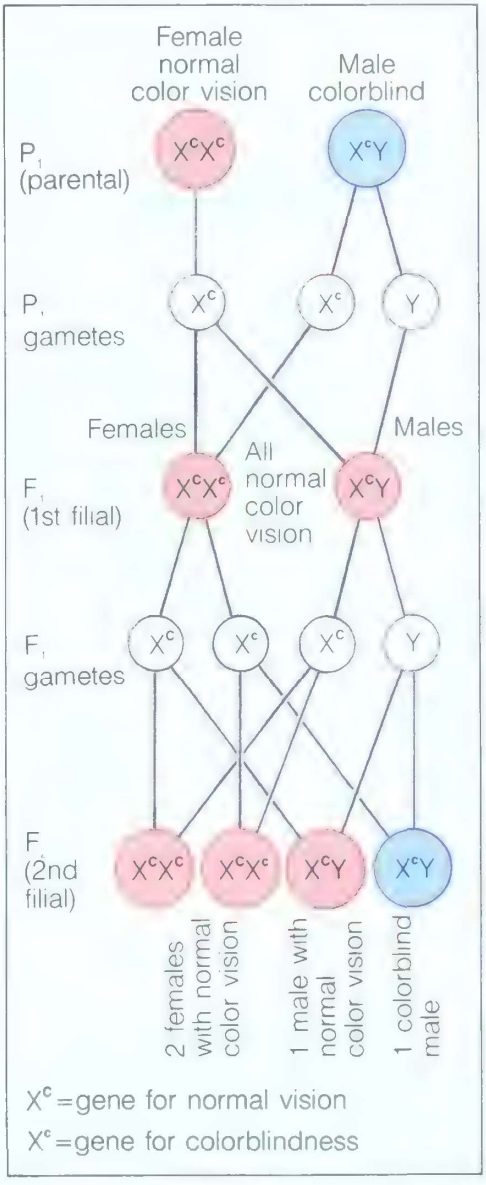
COLORBLINDNESS Colorblindness is a recessive disorder in which a person cannot distinguish between certain colors. **Most types of colorblindness are caused by sex-linked genes located on the X chromosome.** The genes for colorblindness render people unable to make some of the pigments in the eye necessary for color vision.

The most common type of colorblindness is red-green colorblindness. People afflicted with this trait have difficulty distinguishing the lighter shades of red and green. The frequency of this type of colorblindness varies among different populations. In most Caucasian populations, about 8 percent of the males are affected but only about 1 percent of the females are.

In humans, color vision depends on the varying sensitivity of three groups of specialized nerve cells (cones) in the retina of the eye. One group is sensitive to blue light, one to red light, and one to green light. Colors of any given shade excite a specific level of activity from each of the three groups of nerve cells. We will learn more about color vision in Chapter 37.

Because the gene for color vision is carried on the X chromosome, the dominant allele for normal color vision is represented as X^C and the recessive allele for red-green colorblindness is represented as X^c. Homozygous (X^CX^C) and heterozygous (X^CX^c) females have normal color vision. A female who is heterozygous for colorblindness is said to be a carrier because she carries the recessive allele but does not express it. Although she is not colorblind, she is capable of

Figure 11–13 *Colorblindness is an example of a sex-linked trait. In this cross, a female with normal color vision marries a colorblind male (P_1 generation). What are the phenotypes of the offspring in the F_2 generation?*



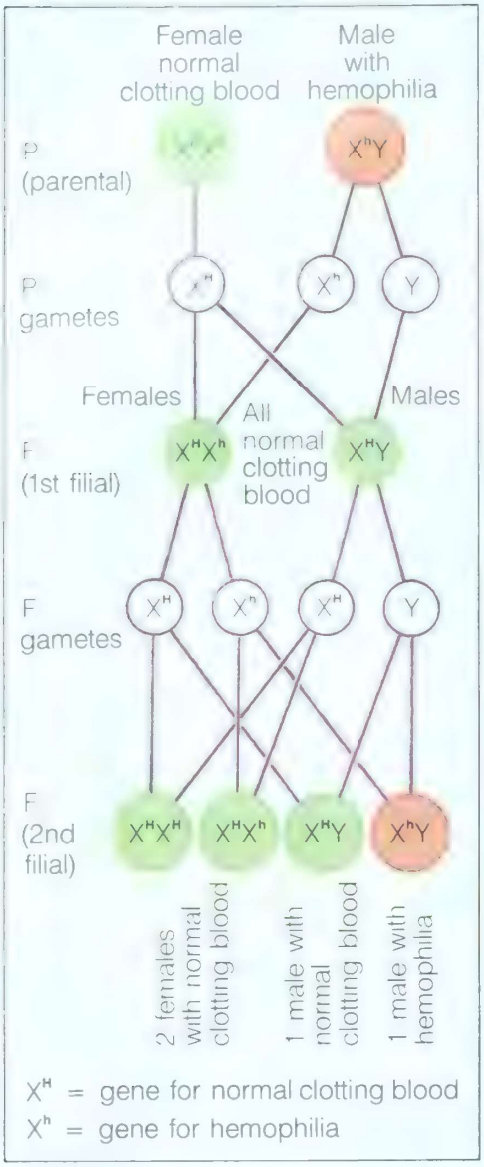


Figure 11–14 *Hemophilia is an example of a sex-linked trait that results in the blood not clotting normally. In this cross, a female with normal-clotting blood marries a male with hemophilia. What are the genotypes of their offspring?*

passing on the gene for colorblindness to her offspring. Only homozygous recessive females (X^cX^c) are colorblind. Because males have only one X chromosome, they are either colorblind (X^cY) or have normal color vision (X^CY).

HEMOPHILIA **Another recessive gene on the X chromosome produces a disorder called hemophilia (hee-moh-FIHL-ee-uh), or "bleeder's disease."** Hemophilia is a disorder in which the protein antihemophilic factor (AHF) necessary for normal blood clotting is missing. Not only is hemophilia a rarer disorder than colorblindness, affecting only about 1 male in 10,000 and only about 1 female in 100,000,000, it is also a more serious disorder.

People who have hemophilia can bleed to death from even the smallest cut. Internal bleeding (hemorrhaging) can occur from the slightest bump or bruise. In recent years, people who have hemophilia have been successfully treated by removing the AHF from donated blood and injecting it into their blood. Although this procedure is inconvenient and dangerous, since it makes people who have hemophilia susceptible to certain diseases carried in the blood, the use of AHF relieves the most serious effects of the disease.

MUSCULAR DYSTROPHY Muscular dystrophy (DIHS-truh-fee) is an inherited disease that results in the progressive wasting away of skeletal muscle. People who have this disease as young children seldom survive past early adulthood. The most common form of this disease, Duchenne muscular dystrophy, is caused by a gene carried on the X chromosome.

Sex-Influenced Traits

Many traits that may seem to be sex-linked, such as male pattern baldness, are actually caused by genes located on autosomes, not on sex chromosomes. Why then is baldness so much more common in men than it is in women? Male pattern



Figure 11–15 *Male pattern baldness is a sex-influenced trait. The expression of this trait is thought to be enhanced by the presence of male sex hormones.*

baldness is a **sex-influenced** trait. **A sex-influenced trait is a trait that is caused by a gene whose expression differs in males and females.**

Many genetic studies indicate that baldness is controlled by a single gene with two alleles, one for normal hair (B) and one for baldness (b). Men and women who have the genotype BB have normal hair, whereas those relatively rare people who have the genotype bb (two baldness alleles) tend to be bald, whether they are male or female. The difference between the sexes occurs in the heterozygous condition, Bb. Males who are heterozygous tend to be bald, whereas females do not. We are not certain how the sex of a person influences the expression of these genes, although it is possible that the male sex hormones may provide an answer.

11–3 SECTION REVIEW

1. How is sex determined in humans?
2. Describe two conditions that are caused by nondisjunction of the sex chromosomes.
3. What are some examples of sex-linked traits?
4. Distinguish between sex-linked and sex-influenced traits.

11–4 Diagnosis of Genetic Disorders

Section Objectives

- Describe how Down syndrome is inherited.
- Identify two methods of detecting genetic disorders during pregnancy.

Humans have been aware of genetic disorders throughout history. For centuries, religious and ethnic laws and customs have reflected a clear understanding on the part of people of the genetic nature of certain disorders. For years, physicians have longed for a way to detect and treat genetic disorders. Today, for some disorders detection is as simple as an examination of a person's chromosomes.

A Chromosomal Abnormality—Down Syndrome

You have just learned about several examples of chromosomal abnormalities. Failures of meiosis, including nondisjunction, cause abnormal combinations of chromosomes to be produced, including those responsible for Turner syndrome and Klinefelter syndrome. Nondisjunction affects autosomes, too.

An example of nondisjunction of autosomes is a condition known as Down syndrome (trisomy 21). In Down syndrome, there is an extra copy of chromosome 21. The presence of this

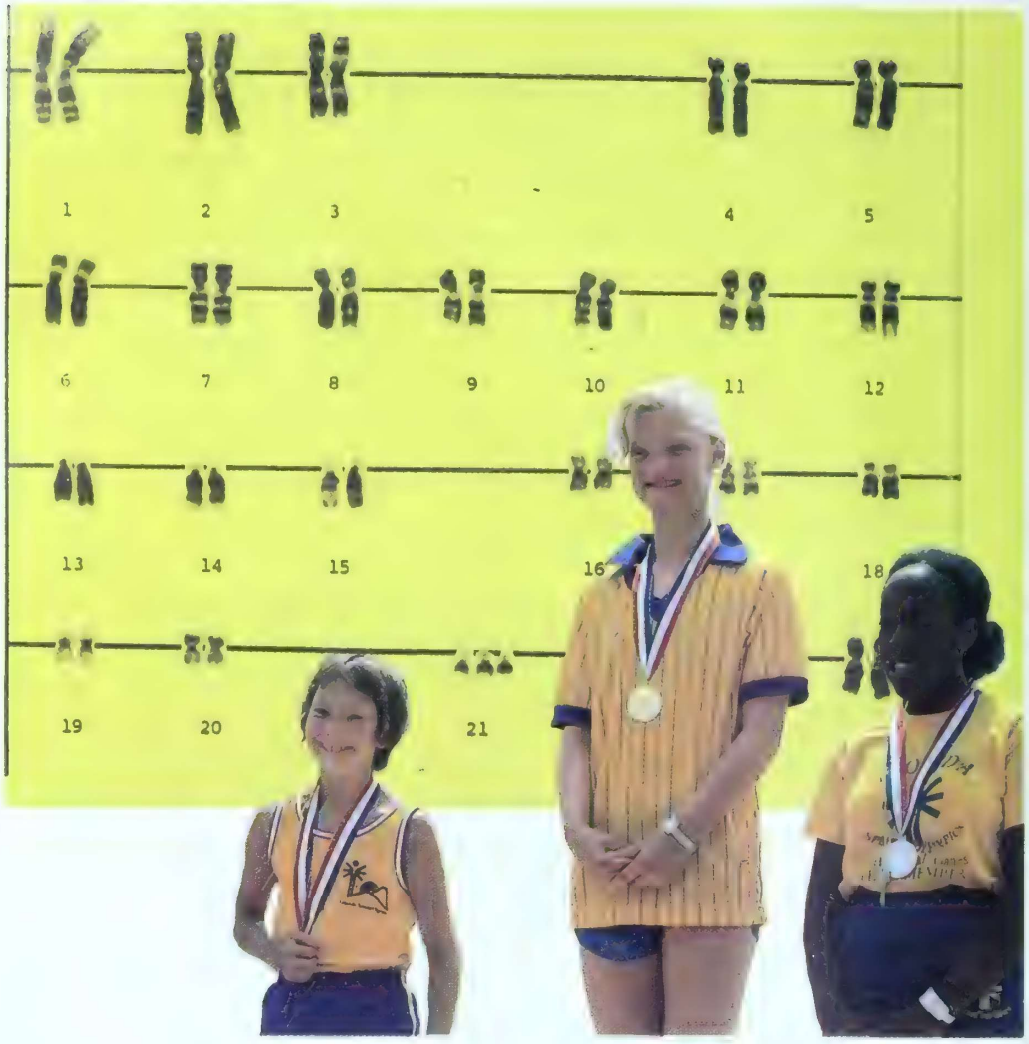


Figure 11–16 *Down syndrome results from the nondisjunction of chromosome 21. Although people with Down syndrome are physically challenged, they can live normal, active lives.*

extra chromosome can be detected in a careful examination of cells under the light microscope.

Down syndrome results in mental retardation that ranges from mild to severe. It is also characterized by an increased susceptibility to many diseases. In the United States, 1 baby in 800 is born with Down syndrome.

It is not clear why an extra copy of one chromosome should cause so much trouble, but scientists have recently discovered rare cases of Down syndrome in which only 46 chromosomes are present. In each of these cases, a small portion of chromosome 21 is attached to another chromosome. Scientists assume that this small portion must contain the genes that cause Down syndrome. A number of other genetic disorders are also produced by chromosomal abnormalities. Like Down syndrome, they too can be diagnosed by microscopic examination of chromosomes.

Prenatal Diagnosis

Down syndrome and other genetic disorders can now be diagnosed before birth by analyzing cells from the developing embryo. One technique, which is known as **amniocentesis**

(am-nee-oh-sehn-TEE-sihs), requires the removal of a small amount of fluid from the sac surrounding the embryo. Cells from the fluid are carefully grown in the laboratory for a few days, treated with a chemical that prevents cell division, and then carefully broken and examined. A karyotype (KAR-ee-uh-tighp), which is a display of all the chromosomes in a cell nucleus, is then prepared to make certain that the chromosomes of the developing embryo are normal.

Chorionic villus biopsy (kor-ee-AHN-ihk VIHL-uhs BIGH-ahp-see), a new alternative to amniocentesis, is a method in which a sample of embryonic cells is removed directly from the membrane surrounding the embryo. Results are obtained more rapidly from this technique than from amniocentesis.

Both amniocentesis and chorionic villus biopsy make it possible to detect Down syndrome and other chromosomal abnormalities. Both techniques are also considered safe for the mother and the developing baby. Scientists are now developing procedures that can detect other genetic disorders. Some of these procedures test for biochemical abnormalities in the embryonic cells, whereas others test for the presence of certain DNA sequences that are characteristic of particular defective genes. The rapid advance of these techniques has made it possible to detect more than 100 genetic disorders from embryonic cells.

Ethical Considerations

Although it is impossible to doubt the effectiveness of prenatal (taking place before birth) diagnosis of genetic disorders, the rapid development of genetic screening techniques is forcing a new set of moral and ethical questions on society. As a society, how should we react to the news that a particular infant will be born with a serious or fatal genetic disorder? Do we favor a society in which such infants are not allowed to develop, or one in which every possible effort is made to preserve all human lives?

Biology has given us great gifts of knowledge and understanding, but it cannot tell us what is proper in such difficult cases. Answers to ethical questions do not come from the laboratory but from the great resources of the human spirit.

11-4 SECTION REVIEW

1. What is Down syndrome?
2. What is amniocentesis? Chorionic villus biopsy?
3. How is a karyotype useful in determining genetic disorders?

LABORATORY INVESTIGATION

OBSERVING HUMAN TRAITS

PROBLEM

What are some patterns of human heredity?

MATERIALS *(per student)*

sheet of paper pencil mirror

PROCEDURE

1. Copy the data table onto the sheet of paper.
2. In the column labeled Trait, circle your phenotype for each trait listed. A description of the dominant allele for each trait follows.
   Tongue rolling (R) is the ability to roll the tongue up at the edges.
   Widow's peak (W) is a hairline that forms a V in the center of the forehead.
   Free earlobes (F) are those that hang below the point of attachment to the head.
   Dimples (D) are indentations on the cheeks.
   Freckles (F) are brownish spots on the skin.
3. Transfer the data from your data table onto the chalkboard, where your teacher has constructed a chart to collect class data.
4. After your classmates have recorded their data on the chalkboard, record the information in the appropriate place in your data table.
5. To determine the percentage of students demonstrating each trait, divide the number of students who have the trait by the total number of students in the class and then multiply this number by 100. Record the data in the appropriate place in your data table.

OBSERVATIONS

1. For each trait, which occurs more frequently: the dominant or recessive allele?
2. Which trait is the most common in your class? The least common?
3. What is the ratio of the percentages for each of the traits?

ANALYSIS AND CONCLUSIONS

1. Do dominant traits occur more often than recessive traits? Explain your answer.
2. What would happen to your results if you were to perform this investigation with five other classes and recorded their data?

Trait		Number of Students Demonstrating Dominant Phenotype	Number of Students Demonstrating Recessive Phenotype	Percentage Demonstrating Dominant Phenotype	Percentage Demonstrating Recessive Phenotype
Dominant	Recessive				
Tongue roller (R)	Nonroller (r)				
Widow's peak (W)	Straight hairline (w)				
Free earlobes (E)	Attached earlobes (e)				
Dimples (D)	No dimples (d)				
Freckles (F)	No freckles (f)				

STUDENT STUDY GUIDE

SUMMARIZING THE CONCEPTS

The key concepts in each section of this chapter are listed below to help you review the chapter content. Make sure you understand each concept and its relationship to other concepts and to the theme of this chapter.

11–1 "It Runs in the Family"

- Many human traits are inherited by the action of dominant and recessive genes, although other traits are determined through more complicated gene interactions.
- Each gamete (sperm cell or egg cell) contains 23 chromosomes.
- During fertilization, sperm and egg unite and a zygote is produced that contains 46 chromosomes.
- Of the 46 chromosomes found in the human diploid cell, 2 are the sex chromosomes (X and Y) and the remaining 44 are called autosomes.

11–2 The Inheritance of Human Traits

- Multiple alleles are three or more alleles of the same gene that code for a single trait. ABO and Rh blood groups are determined by multiple alleles.
- Sickle cell anemia is a disorder that causes the normally disk-shaped red blood cells to become sickle-shaped. The gene for normal hemoglobin is codominant with the sickle cell gene.
- Polygenic traits are traits that are controlled by a number of genes.

11–3 Sex-Linked Inheritance

- Nondisjunction is the failure of chromosomes to separate during one of the stages of meiosis.
- Genes that are carried on either the X or the Y chromosome are said to be sex-linked.
- A sex-influenced trait is a trait that is caused by a gene whose expression differs in males and females.

11–4 Diagnosis of Genetic Disorders

- Down syndrome is an example of a condition caused by the nondisjunction of autosomes.
- Many genetic disorders can now be diagnosed before birth because of such genetic screening techniques as amniocentesis and chorionic villus biopsy.

REVIEWING KEY TERMS

Vocabulary terms are important to your understanding of biology. The key terms listed below are those you should be especially familiar with. Review these terms and their meanings. Then use each term in a complete sentence. If you are not sure of a term's meaning, return to the appropriate section and review its definition.

11–1 "It Runs in the Family"	11–2 The Inheritance of Human Traits	11–3 Sex-Linked Inheritance	11–4 Diagnosis of Genetic Disorders
gamete	multiple allele	sex-linked	amniocentesis
zygote	polygenic trait	sex-influenced	chorionic villus biopsy

CHAPTER REVIEW

CONTENT REVIEW

Multiple Choice

Choose the letter of the answer that best completes each statement.

1. An example of a trait that is determined by multiple alleles is
   a. Huntington disease.
   b. ABO blood groups.
   c. Down syndrome.
   d. hemophilia.
2. Three brothers have blood types A, B, and O. What are the chances that the parents of these three will produce a fourth child whose blood type is AB?
   a. 0 percent
   b. 25 percent
   c. 50 percent
   d. 100 percent
3. A disorder that results from a change in an amino acid in the hemoglobin molecule is
   a. sickle cell anemia.
   b. hemophilia.
   c. Down syndrome.
   d. Turner syndrome.
4. Which is an example of a polygenic trait?
   a. skin color
   b. hemophilia
   c. ABO blood groups
   d. Huntington disease
5. Which parental pair could produce a colorblind female?
   a. homozygous normal vision mother and colorblind father
   b. colorblind mother and normal vision father
   c. heterozygous normal vision mother and normal vision father
   d. heterozygous normal vision mother and colorblind father
6. Which is an example of a sex-influenced trait?
   a. Down syndrome
   b. colorblindness
   c. male pattern baldness
   d. skin color
7. What is the total number of chromosomes in a diploid cell of a person with Down syndrome?
   a. 22
   b. 23
   c. 44
   d. 47
8. Which of the following techniques is directly associated with the diagnoses of certain genetic disorders by the examination of chromosomes?
   a. blood typing
   b. karyotyping
   c. chemical screening
   d. microsurgery

True or False

Determine whether each statement is true or false. If it is true, write "true." If it is false, change the underlined word or words to make the statement true.

1. During fertilization, sperm and egg unite and produce a gamete.
2. A person with type A blood has red blood cells that contain antigen B.
3. A genotype Rh^+Rh^- will produce the Rh negative phenotype.
4. People who are heterozygous for sickle cell anemia are partially resistant to malaria.
5. Based on the pattern of inheritance of sex-linked traits, if a male has hemophilia, he has one gene for this trait on the sex chromosomes in each of his diploid cells.
6. The dominant allele for normal color vision is represented as X^c.
7. In Down syndrome, there is an extra copy of chromosome 16.
8. Chorionic villus biopsy is a genetic screening technique in which a small amount of fluid is removed from the sac surrounding the developing embryo.

CONCEPT MASTERY

Use your understanding of the concepts developed in the chapter to answer each of the following in a brief paragraph.

1. Why are humans not considered good organisms for genetic studies?
2. Explain why type O blood is sometimes referred to as the universal donor. Is this true in the case of O^+ blood?
3. What is the cause of sickle cell anemia?
4. Why is it easy to believe that the sickle cell trait may have arisen independently in several places throughout the world?
5. What is the evolutionary connection between the incidences of sickle cell anemia and the incidences of malaria?
6. Why are there more colorblind males than there are colorblind females?
7. What evidence suggests that an X chromosome is essential for normal development? Is the same true of the Y chromosome?
8. The genes for red-green colorblindness and hemophilia are located on the X chromosome. How might you determine whether these genes are closely linked?
9. How would you determine whether a human gene was sex-linked?

CRITICAL AND CREATIVE THINKING

Discuss each of the following in a brief paragraph.

1. **Applying concepts** Cystic fibrosis (CF) is caused by an autosomal recessive gene and is a fatal disorder that affects people of European ancestry. CF is characterized by a production of thick mucus that clogs the lungs and parts of the digestive system, making it difficult to breathe or eat. Suppose two people without a history of CF in their immediate families have a child with CF. They would like to have another child, but they are concerned about whether the second child will have CF. Can you help them predict the chances of having a second child with CF?
2. **Making diagrams** Polydactyly is a human characteristic in which a person has extra digits (fingers or toes) on his or her hands or feet. The trait for polydactyly is dominant over the trait for five digits on the hands and/or feet. Suppose a man who is heterozygous for this trait marries a woman with the normal number of digits. What are the possible genotypes and phenotypes of their offspring? Draw a Punnett square showing the possible results of this cross.
3. **Relating concepts** Nine of ten children in a family are right-handed. Right-handedness is a dominant trait. Assume that this trait is controlled by a pair of genes.
   a. What is the genotype of a left-handed child?
   b. What are the possible genotypes of the right-handed children?
   c. What are the possible genotypes of the parents?
   d. Could these parents have produced only right-handed children? Explain.
   e. If these parents had produced only one child, would this child have been left- or right-handed? Explain your answer.
4. **Making inferences** Discuss the impact of genetic screening techniques on society.
5. **Using the writing process** Your career ambition is to be a science reporter. You are sent by your school newspaper to interview a physician who works with human genetic disorders. Prepare a script of the questions you would like answered.