Mendelian Genetics

10

CHAPTER 10

Chapter Outline

Key Concepts	Applications
Understand the concepts of genotype and phenotype.	• Explain how a person can have the allele for a particular trait but not show it.
Understand the basics of Mendelian genetics.	• Determine if the children of a father and a mother with a certain gene combination will automatically show that trait. • Understand how people inherit varying degrees of traits such as skin color.
Work single-gene and double-factor genetic problems.	• Determine the likelihood that a particular trait will be passed on to the next generation. • Determine the chances that children will carry two particular genes.
Understand how a person's sex can influence the expression of their genes.	• Explain why men and women inherit some traits differently.
Understand how genes and their alleles interact.	• Use the concepts of dominant alleles and recessive alleles, incompletely dominant alleles, and X-linkage to explain inheritance patterns.

10.1 Genetics, Meiosis, and Cells

Why do you have a particular blood type or hair color? Why do some people have the same skin color as their parents and others have a skin color different from that of their parents? Why do flowers show such a wide variety of colors? Why is it that generation after generation of plants, animals, and microbes look so much like members of their own kind? These questions and many others can be better answered if you have an understanding of genetics.

A **gene** is a portion of DNA that determines a characteristic. Through meiosis and reproduction, genes can be transmitted from one generation to another. The study of genes, how genes produce characteristics, and how the characteristics are inherited is the field of biology called **genetics.** The first person to systematically study inheritance and formulate laws about how characteristics are passed from one generation to the next was an Augustinian monk named Gregor Mendel (1822–1884). Mendel's work was not generally accepted until 1900, when three men, working independently, rediscovered some of the ideas that Mendel had formulated more than 30 years earlier. Because of his early work, the study of the pattern of inheritance that follows the laws formulated by Gregor Mendel is often called **Mendelian genetics.**

To understand this chapter, you need to know some basic terminology. One term that you have already encountered is *gene*. Mendel thought of a gene as a *particle* that could be passed from the parents to the **offspring** (*children*, *descendants*, or *progeny*). Today we know that genes are actually composed of specific sequences of DNA nucleotides. The particle concept is not entirely inaccurate, because a particular gene is located at a specific place on a chromosome called its **locus** (*locus* = location; plural, loci).

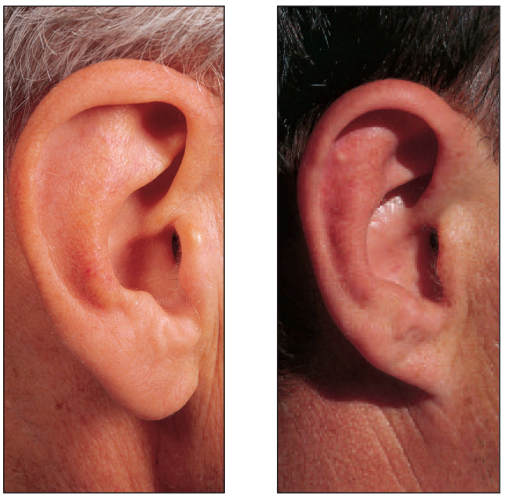
Another important idea to remember is that all sexually reproducing organisms have a diploid ($2n$) stage. Because gametes are haploid (n) and most organisms are diploid, the conversion of diploid to haploid cells during meiosis is an important process.

$$\mathbf{2(}n\mathbf{)} \rightarrow \textbf{meiosis} \rightarrow \mathbf{(}n\mathbf{)}\ \textbf{gametes}$$

The diploid cells have two sets of chromosomes—one set inherited from each parent.

$$n + n\ \textbf{gametes} \rightarrow \textbf{fertilization} \rightarrow 2n$$

Therefore, they have two chromosomes of each kind and have two genes for each characteristic. When sex cells are produced by meiosis, reduction division occurs, and the diploid number is reduced to haploid. Therefore, the sex cells produced by meiosis have only one chromosome of each of the homologous pairs that were in the diploid cell that began meiosis. Diploid organisms usually result from the fertilization of a haploid egg by a haploid sperm. Thus they inherit one gene of each type from each parent. For example, each of us has two genes for earlobe shape: one came with our father's sperm, the other with our mother's egg (figure 10.1).



(a) (b)

Figure 10.1

Genes Control Structural Features
Whether your earlobe is free (*a*) or attached (*b*) depends on the genes you have inherited. As genes express themselves, their actions affect the development of various tissues and organs. Some people's earlobes do not separate from the sides of their heads in the same manner as do those of others. How genes control this complex growth pattern and why certain genes function differently than others is yet to be clarified.

10.2 Single-Gene Inheritance Patterns

In diploid organisms there may be two different forms of the gene. In fact, there may be *several* alternative forms or **alleles** of each gene within a population. In people, for example, there are two alleles for earlobe shape. One allele produces an earlobe that is fleshy and hangs free, whereas the other allele produces a lobe that is attached to the side of the face and does not hang free. The type of earlobe that is present is determined by the type of allele (gene) received from each parent and the way in which these alleles interact with one another. Alleles are located on the same pair of homologous chromosomes—one allele on each chromosome. These alleles are also at the same specific location, or locus (figure 10.2).

The **genome** is a set of all the genes necessary to specify an organism's complete list of characteristics. The term genome is used in two ways. It may refer to the diploid ($2n$) or haploid (n) number of chromosomes in a cell. Be sure to clarify how this term is used by your instructor. The **genotype** of an organism is a listing of the genes present in that organism. It consists of the cell's DNA code; therefore, you cannot see the genotype of an organism. It is not yet possible to know the complete genotype of most organisms, but it is often possible to figure out the genes present that determine a particular characteristic. For example, there are three possible

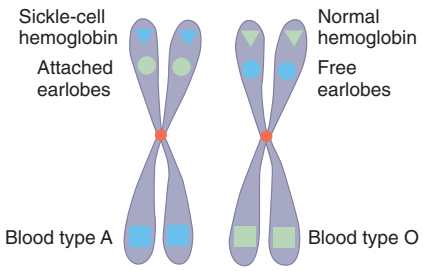


Figure 10.2

A Pair of Homologous Chromosomes
Homologous chromosomes contain genes for the same characteristics at the same place. Note that the attached-earlobe allele is located at the ear-shape locus on one chromosome, and the free-earlobe allele is located at the ear-shape locus on the other member of the homologous pair of chromosomes. The other two genes are for hemoglobin structure (alleles for normal and sickled cells) and blood type (alleles for blood types A and O). The examples presented here are for illustrative purposes only. We do not really know if these particular genes are on these chromosomes. It is hoped that the Human Genome Project, described in chapter 9, will resolve this problem.

genotypic combinations of the two alleles for earlobe shape. Genotypes are typically represented by upper- and lowercase letters. In the case of the earlobe trait, the allele for free earlobes is designated "*E*," whereas that for attached earlobes is "*e*." A person's genotype could be (1) two alleles for attached earlobes, (2) one allele for attached earlobes and one allele for free earlobes, or (3) two alleles for free earlobes.

How would individuals with each of these three genotypes look? The way each combination of alleles *expresses* (shows) itself is known as the **phenotype** of the organism. The phrase **gene expression** refers to the degree to which a gene goes through transcription and translation to show itself as an observable feature of the individual.

A person with two alleles for attached earlobes will have earlobes that do not hang free. A person with one allele for attached earlobes and one allele for free earlobes will have a phenotype that exhibits free earlobes. An individual with two alleles for free earlobes will also have free earlobes. Notice that there are three genotypes, but only two phenotypes. The individuals with the free-earlobe phenotype can have different genotypes.

Alleles	*Genotypes*	*Phenotypes*
E = free earlobes	*EE*	Free earlobes
e = attached earlobes	*Ee*	Free earlobes
	ee	Attached earlobes

The expression of some genes is directly influenced by the presence of other alleles. For any particular pair of alleles in an individual, the two alleles from the two parents are either identical or not identical. Persons are **homozygous** for a trait when they have the combination of two identical alleles for that particular characteristic, for example, *EE* and *ee*. A person with two alleles for freckles is said to be homozygous for that trait. A person with two alleles for no freckles is also homozygous. If an organism is homozygous, the characteristic expresses itself in a specific manner. A person homozygous for free earlobes has free earlobes, and a person homozygous for attached earlobes has attached earlobes.

Individuals are designated as **heterozygous** when they have two different allelic forms of a particular gene, for example, *Ee*. The heterozygous individual received one form of the gene from one parent and a different allele from the other parent. For instance, a person with one allele for freckles and one allele for no freckles is heterozygous. If an organism is heterozygous, these two different alleles interact to determine a characteristic. A **carrier** is any person who is heterozygous for a trait. In this situation, the recessive allele is hidden, that is, does not express itself enough to be a phenotype.

Dominant and Recessive Alleles

Often, one allele in the pair expresses itself more than the other. A **dominant allele** masks the effect of other alleles for the trait. For example, if a person has one allele for free earlobes and one allele for attached earlobes, that person has a phenotype of free earlobes. We say the allele for free earlobes is dominant. A **recessive allele** is one that, when present with another allele, has its actions overshadowed by the other; it is masked by the effect of the other allele. Having attached earlobes is the result of having a combination of two recessive characteristics. A person with one allele for free earlobes and one allele for attached earlobes has a phenotype of free earlobes. The expression of recessive alleles is only noted when the organism is homozygous for the recessive alleles. If you have attached earlobes, you have two alleles for that trait. Don't think that recessive alleles are necessarily bad. *The term recessive has nothing to do with the significance or value of the allele—it simply describes how it can be expressed. Recessive alleles are not less likely to be inherited but must be present in a homozygous condition to express themselves. Also, recessive alleles are not necessarily less frequent in the population* (see table 11.1). Sometimes the physical environment determines whether or not dominant or recessive genes function. For example, in humans genes for freckles do not show themselves fully unless a person's skin is exposed to sunlight (figure 10.3).

Codominance

In cases of dominance and recessiveness, one allele of the pair clearly overpowers the other. Although this is common, it is not always the case. In some combinations of alleles, there is a **codominance.** This is a situation in which both alleles in a heterozygous condition express themselves.

Figure 10.3

The Environment and Gene Expression
The expression of many genes is influenced by the environment. The allele for dark hair in the cat is sensitive to temperature and expresses itself only in the parts of the body that stay cool. The allele for freckles expresses itself more fully when a person is exposed to sunlight.

A classic example of codominance in plants involves the color of the petals of snapdragons. There are two alleles for the color of these flowers. Because neither allele is recessive, we cannot use the traditional capital and small letters as symbols for these alleles. Instead, the allele for white petals is given the symbol F^W, and the one for red petals is given the symbol F^R (figure 10.4). There are three possible combinations of these two alleles:

Genotype	*Phenotype*
F^WF^W	White flower
F^RF^R	Red flower
F^RF^W	Pink flower

Notice that there are only two different alleles, red and white, but there are three phenotypes, red, white, and pink. Both the red-flower allele and the white-flower allele partially express themselves when both are present, and this results in pink.

A human example involves the genetic abnormality, *sickle-cell disease* (see figure 7.11). Having the two recessive alleles for sickle-cell hemoglobin (Hb^S and Hb^S) can result in abnormally shaped red blood cells. This occurs because the hemoglobin molecules are synthesized with the wrong amino acid sequence. These abnormal hemoglobin molecules tend to attach to one another in long, rodlike chains when oxygen is in short supply, that is, with exercise, pneumonia, emphysema. These rodlike chains distort the shape of the red blood cells into a sickle shape. When these abnormal red blood cells change shape, they clog small blood vessels. The sickled red cells are also destroyed more rapidly than normal cells. This results in a shortage of red blood cells, a condition known as anemia, and an oxygen deficiency in the tissues that have become clogged. People with sickle-cell anemia may experience pain, swelling, and damage to organs such as the heart, lungs, brain, and kidneys.

Figure 10.4

A Case of Codominance
The colors of these snapdragons are determined by two alleles for petal color, F^W and F^R. There are three different phenotypes because of the way in which the alleles interact with one another. In the heterozygous condition, neither of the alleles dominates the other.

Sickle-cell anemia can be lethal in the homozygous recessive condition. In the homozygous dominant condition (Hb^AHb^A), the person has normal red blood cells. In the heterozygous condition (Hb^AHb^S), patients produce both kinds of red blood cells. When the amount of oxygen in the blood falls below a certain level, those able to sickle will distort. However, when this occurs, most people heterozygous for the trait do not show severe symptoms. Therefore these alleles are related to one another in a codominant fashion. However, under the right circumstances, being heterozygous can be beneficial. A person with a single sickle-cell allele is more resistant to malaria than a person without this allele.

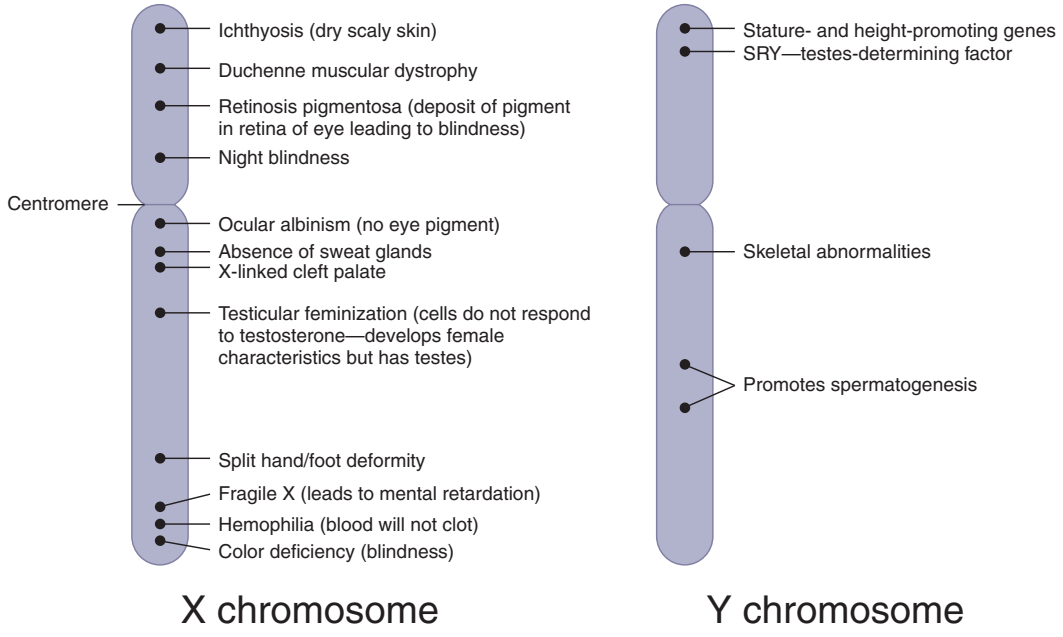


Figure 10.5

Sex Chromosomes
Why is the Y chromosome so small? Is there an advantage to a species in having one sex chromosome deficient in genes? One hypothesis answers yes! Consider the idea that, with genes for supposedly "female" characteristics eliminated from the Y chromosome, crossing-over and recombining with "female" genes on the X chromosome during meiosis could help keep sex traits separated. Males would be males and females would stay females. The chances of "male-determining" and "female-determining" genes getting mixed onto the same chromosome would be next to impossible because they would not even exist on the Y chromosome.

Genotype	*Phenotype*
$Hb^A\ Hb^A$	**Normal hemoglobin and nonresistance to malaria**
$Hb^A\ Hb^S$	**Normal hemoglobin and resistance to malaria**
$Hb^S\ Hb^S$	**Resistance to malaria but death from sickle-cell anemia**

Originally, sickle-cell anemia was found at a high frequency in parts of the world where malaria was common, such as tropical regions of Africa and South America. Today, however, this genetic disease can be found anywhere in the world. In the United States, it is most common among black populations whose ancestors came from equatorial Africa.

X-Linked Genes

Pairs of alleles located on nonhomologous chromosomes separate independently of one another during meiosis when the chromosomes separate into sex cells. Because each chromosome has many genes on it, these genes tend to be inherited as a group. Genes located on the same chromosome that tend to be inherited together are called a **linkage group.** The process of crossing-over, which occurs during prophase I of meiosis I, may split up these linkage groups. Crossing-over happens between homologous chromosomes donated by the mother and the father and results in a mixing of genes. The closer two genes are to each other on a chromosome, the more probable it is that they will be inherited together.

People and many other organisms have two types of chromosomes. **Autosomes** (22 pairs) are not involved in sex determination and have the same kinds of genes on both members of the homologous pair of chromosomes. **Sex chromosomes** are a pair of chromosomes that control the sex of an organism. In humans, and some other animals, there are two types of sex chromosomes—the X chromosome and the Y chromosome. The Y chromosome is much shorter than the X chromosome and has fewer genes for traits than found on the X chromosome (figure 10.5). One genetic trait that is located on the Y chromosome contains the testis-determining gene—SRY. Females are normally produced when two X chromosomes are present. Males are usually produced when one X chromosome and one Y chromosome are present.

Genes found together on the X chromosome are said to be **X-linked.** Because the Y chromosome is shorter than the X chromosome, it does not have many of the alleles that are found on the comparable portion of the X chromosome. Therefore, in a man, the presence of a single allele on his

only X chromosome will be expressed, regardless of whether it is dominant or recessive. A Y-linked trait in humans is the SRY gene. This gene controls the differentiation of the embryonic gonad to a male testis. By contrast, more than 100 genes are on the X chromosome. Some of these X-linked genes can result in abnormal traits such as *color deficiency, hemophilia, brown teeth,* and at least two forms of *muscular dystrophy* (Becker's and Duchenne's).

10.3 Mendel's Laws of Heredity

Heredity problems are concerned with determining which alleles are passed from the parents to the offspring and how likely it is that various types of offspring will be produced. The first person to develop a method of predicting the outcome of inheritance patterns was Mendel, who performed experiments concerning the inheritance of certain characteristics in garden pea (*pisum satium*) plants. From his work, Mendel concluded which traits were dominant and which were recessive. Some of his results are shown in table 10.1.

What made Mendel's work unique was that he studied only one trait at a time. Previous investigators had tried to follow numerous traits at the same time. When this was attempted, the total set of characteristics was so cumbersome to work with that no clear idea could be formed of how the offspring inherited traits. Mendel used traits with clear-cut alternatives, such as purple or white flower color, yellow or green seed pods, and tall or dwarf pea plants. He was very lucky to have chosen pea plants in his study because they naturally self-pollinate. When self-pollination occurs in pea plants over many generations, it is possible to develop a population of plants that is homozygous for a number of characteristics. Such a population is known as a *pure line.*

Mendel took a pure line of pea plants having purple flower color, removed the male parts (anthers), and discarded them so that the plants could not self-pollinate. He then took anthers from a pure-breeding white-flowered plant and pollinated the antherless purple flower. When the pollinated flowers produced seeds, Mendel collected, labeled, and planted them. When these seeds germinated and grew, they eventually produced flowers.

You might be surprised to learn that all the plants resulting from this cross had purple flowers. One of the prevailing hypotheses of Mendel's day would have predicted that the purple and white colors would have blended, resulting in flowers that were lighter than the parental purple flowers. Another hypothesis would have predicted that the offspring would have had a mixture of white and purple flowers. The unexpected result—all the offspring produced flowers like those of one parent and no flowers like those of the other—caused Mendel to examine other traits as well and formed the basis for much of the rest of his work. He repeated his experiments using pure strains for other traits. Pure-breeding tall plants were crossed with pure-breeding dwarf plants. Pure-breeding plants with yellow pods were crossed with pure-breeding plants with green pods. The results were all the same: the offspring showed the characteristics of one parent and not the other.

Table 10.1

DOMINANT AND RECESSIVE TRAITS IN PEA PLANTS

Characteristic	Dominant Allele	Recessive Allele
Plant height	Tall	Dwarf
Pod shape	Full	Constricted
Pod color	Green	Yellow
Seed surface	Round	Wrinkled
Seed color	Yellow	Green
Flower color	Purple	White

Next, Mendel crossed the offspring of the white-purple cross (all of which had purple flowers) with each other to see what the third generation would be like. Had the characteristic of the original white-flowered parent been lost completely? This second-generation cross was made by pollinating these purple flowers that had one white parent among themselves. The seeds produced from this cross were collected and grown. When these plants flowered, three-fourths of them produced purple flowers and one-fourth produced white flowers.

After analyzing his data, Mendel formulated several genetic laws to describe how characteristics are passed from one generation to the next and how they are expressed in an individual.

Mendel's **law of dominance** When an organism has two different alleles for a given trait, the allele that is expressed, overshadowing the expression of the other allele, is said to be *dominant.* The gene whose expression is overshadowed is said to be *recessive.*

Mendel's **law of segregation** When gametes are formed by a diploid organism, the alleles that control a trait separate from one another into different gametes, retaining their individuality.

Mendel's **law of independent assortment** Members of one gene pair separate from each other independently of the members of other gene pairs.

At the time of Mendel's research, biologists knew nothing of chromosomes or DNA or of the processes of mitosis and meiosis. Mendel assumed that each gene was separate from other genes. It was fortunate for him that most of the characteristics he picked to study were found on separate chromosomes. If two or more of these genes had been located on the same chromosome (*linked genes*), he probably would not have been able to formulate his laws. The discovery of chromosomes and DNA have led to modifications in Mendel's laws, but it was Mendel's work that formed the foundation for the science of genetics.

10.4 Probability Versus Possibility

In order to solve heredity problems, you must have an understanding of probability. **Probability** is the chance that an event will happen, and is often expressed as a percentage or a fraction. *Probability* is not the same as *possibility*. It is possible to toss a coin and have it come up heads. But the probability of getting a head is more precise than just saying it is possible to get a head. The probability of getting a head is 1 out of 2 (½ or 0.5 or 50%) because there are two sides to the coin, only one of which is a head. Probability can be expressed as a fraction:

$$\text{Probability} = \frac{\text{the number of events that can produce a given outcome}}{\text{the total number of possible outcomes}}$$

What is the probability of cutting a deck of cards and getting the ace of hearts? The number of times that the ace of hearts can occur is 1. The total number of possible outcomes (number of cards in the deck) is 52. Therefore, the probability of cutting an ace of hearts is 1/52.

What is the probability of cutting an ace? The total number of aces in the deck is 4, and the total number of cards is 52. Therefore, the probability of cutting an ace is 4/52 or 1/13.

It is also possible to determine the probability of two independent events occurring together. *The probability of two or more events occurring simultaneously is the product of their individual probabilities.* If you throw a pair of dice, it is possible that both will be 4s. What is the probability that both will be 4s? The probability of one die being a 4 is ⅙. The probability of the other die being a 4 is also ⅙. Therefore, the probability of throwing two 4s is

$$1/6 \times 1/6 = 1/36$$

10.5 Steps in Solving Heredity Problems: Single-Factor Crosses

The first type of problem we will consider is the easiest type, a single-factor cross. A **single-factor cross** (sometimes called a monohybrid cross: *mono* = one; *hybrid* = combination) is a genetic cross or mating in which a single characteristic is followed from one generation to the next. For example, in humans, the allele for *Tourette syndrome (TS)* is inherited as an autosomal dominant allele.

For centuries, people displaying this genetic disorder were thought to be possessed by the devil since they displayed such unusual behaviors. These motor and verbal behaviors or *tics* are involuntary and range from mild (e.g., leg tapping, eye blinking, face twitching) to the more violent forms such as the shouting of profanities, head jerking, spitting, compulsive repetition of words, or even barking like a dog. The symptoms result from an excess production of the brain messenger, dopamine.

If both parents are heterozygous (have one allele for Tourette and one allele for no Tourette syndrome) what is the probability that they can have a child without Tourette syndrome? With Tourette syndrome?

Steps in Solving Heredity Problems—Single-Factor Crosses

Five basic steps are involved in solving a heredity problem.

Step 1: Assign a Symbol for Each Allele.

Usually a capital letter is used for a dominant allele and a small letter for a recessive allele. Use the symbol *T* for Tourette and *t* for no Tourette.

Allele	*Genotype*	*Phenotype*
T = Tourette	*TT*	Tourette syndrome
t = normal	*Tt*	Tourette syndrome
	tt	Normal

Step 2: Determine the Genotype of Each Parent and Indicate a Mating.

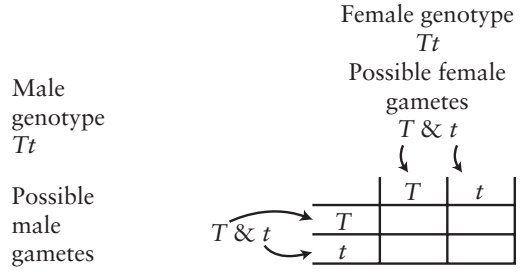
Because both parents are heterozygous, the male genotype is *Tt*. The female genotype is also *Tt*. The × between them is used to indicate a mating.

$$Tt \times Tt$$

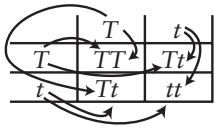
Step 3: Determine All the Possible Kinds of Gametes Each Parent Can Produce.

Remember that gametes are haploid; therefore, they can have only one allele instead of the two present in the diploid cell. Because the male has both the Tourette syndrome allele and the normal allele, half his gametes will contain the Tourette syndrome allele and the other half will contain the normal allele. Because the female has the same genotype, her gametes will be the same as his.

For genetic problems, a *Punnett square* is used. A **Punnett square** is a box figure that allows you to determine the probability of genotypes and phenotypes of the progeny of a particular cross. Remember, because of the process of meiosis, each gamete receives only one allele for each characteristic listed. Therefore, the male will produce sperm with either a *T* or a *t;* the female will produce ova with either a *T* or a *t*. The possible gametes produced by the male parent are listed on the left side of the square and the female gametes are listed on the top. In our example, the Punnett square would show a single dominant allele and a single recessive allele from the male on the left side. The alleles from the female would appear on the top.



Step 4: Determine All the Gene Combinations That Can Result When These Gametes Unite.
To determine the possible combinations of alleles that could occur as a result of this mating, simply fill in each of the empty squares with the alleles that can be donated from each parent. Determine all the gene combinations that can result when these gametes unite.



Step 5: Determine the Phenotype of Each Possible Gene Combination.
In this problem, three of the offspring, *TT*, *Tt*, and *Tt*, have Tourette syndrome. One progeny, *tt*, is normal. Therefore, the answer to the problem is that the probability of having offspring with Tourette syndrome is ¾; for no Tourette syndrome, it is ¼.

Take the time to learn these five steps. All single-factor problems can be solved using this method; the only variation in the problems will be the types of alleles and the number of possible types of gametes the parents can produce. Now let's consider a problem in which one parent is heterozygous and the other is homozygous for a trait.

Problem: Dominant/Recessive PKU
Some people are unable to convert the amino acid phenylalanine into the amino acid tyrosine. The buildup of phenylalanine in the body prevents the normal development of the nervous system. Such individuals suffer from phenylketonuria (PKU) and may become mentally retarded (figure 10.6). The normal condition is to convert phenylalanine to tyrosine. It is dominant over the condition for PKU. If one parent is heterozygous and the other parent is homozygous for PKU, what is the probability that they will have a child who is normal? A child with PKU?

Step 1:
Use the symbol *N* for normal and *n* for PKU.

Allele	*Genotype*	*Phenotype*
***N* = normal**	***NN***	**Normal metabolism of phenylalanine**
***n* = PKU**	***Nn***	**Normal metabolism of phenylalanine**
	nn	**PKU disorder**

Step 2:

$$Nn \times nn$$

Step 3:

	n
N	
n	

Step 4:

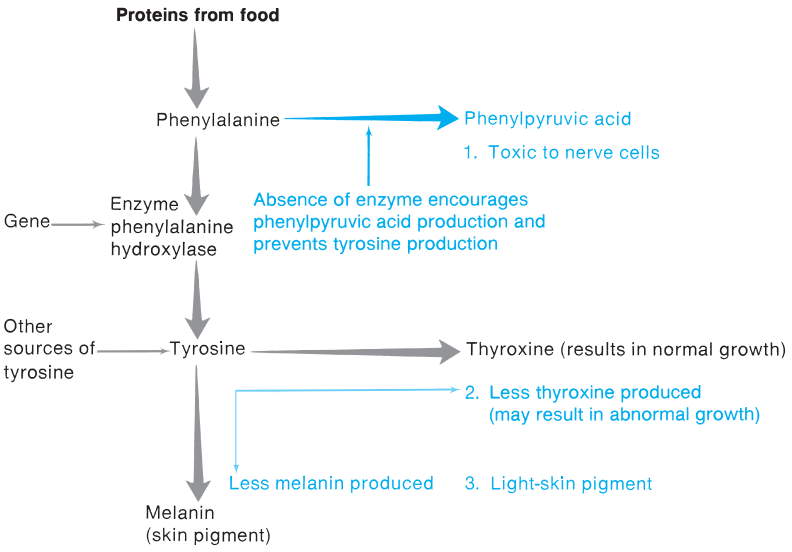
	n
N	***Nn***
n	***nn***

Step 5:
In this problem, ½ of the progeny will be normal and ½ will have PKU.

Problem: Codominance
If a pink snapdragon is crossed with a white snapdragon, what phenotypes can result, and what is the probability of each phenotype?

Figure 10.6

Phenylketonuria
PKU is an autosomal recessive disorder located on chromosome 12. This diagram shows how the normal pathways work (these are shown in gray). If the enzyme phenylalanine hydroxylase is not produced because of a mutated gene, the amino acid phenylalanine cannot be broken down, and is converted into phenylpyruvic acid which accumulates in body fluids. There are three major results: (*1*) mental retardation because phenylpyruvic acid kills nerve cells, (*2*) abnormal body growth because less of the growth hormone thyroxine is produced, and (*3*) pale skin pigmentation because less melanin is produced (abnormalities are shown in color). It should also be noted that if a woman who has PKU becomes pregnant, her baby is likely to be born retarded. Although the embryo may not have the genetic disorder, the phenylpyruvic acid produced by the pregnant mother will damage the developing brain cells. This is called *maternal PKU.*



Step 1:

F^W = white flowers F^R = red flowers

Genotype	*Phenotype*
F^WF^W	White flower
F^WF^R	Pink flower
F^RF^R	Red flower

Step 2:

$$F^RF^W \times F^WF^W$$

Step 3:

	F^W
F^R	
F^W	

Step 4:

	F^W
F^R	F^WF^R Pink flower
F^W	F^WF^W White flower

Step 5:

This cross results in two different phenotypes—pink and white. No red flowers can result because this would require that both parents be able to contribute at least one red allele. The white flowers are homozygous for white, and the pink flowers are heterozygous.

Problem: X-Linked

In humans, the gene for normal color vision is dominant and the gene for color deficiency is recessive. Both genes are X-linked. People who are color blind are not really blind, but should more appropriately be described as having "color defective vision." A male who has normal vision mates with a female who is heterozygous for normal color vision. What type of children can they have in terms of these traits, and what is the probability for each type?

Step 1:

This condition is linked to the X chromosome, so it has become traditional to symbolize the allele as a superscript on the letter X. Because the Y chromosome does not contain a homologous allele, only the letter Y is used.

X^N = normal color vision
X^n = color-deficient
Y = male (no gene present)

Genotype	*Phenotype*
X^NY	Male, normal color vision
X^nY	Male, color-deficient
X^NX^N	Female, normal color vision
X^NX^n	Female, normal color vision
X^nX^n	Female, color-deficient

Step 2:

Male's genotype = X^NY (normal color vision)
Female's genotype = X^NX^n (normal color vision)

$$X^NY \times X^NX^n$$

Step 3:

The genotype of the gametes are listed in the Punnett square:

	X^N	X^n
X^N		
Y		

Step 4:

The genotypes of the probable offspring are listed in the body of the Punnett square:

	X^N	X^n
X^N	X^NX^N	X^NX^n
Y	X^NY	X^nY

Step 5:

The phenotypes of the offspring are determined:

Normal female	Carrier female
Normal male	Color-deficient male

10.6 The Double-Factor Cross

A **double-factor cross** is a genetic study in which two pairs of alleles are followed from the parental generation to the offspring. Sometimes this type of cross is referred to as a dihybrid (*di* = two; *hybrid* = combination) cross. This problem is solved in basically the same way as a single-factor cross. The main difference is that in a double-factor cross you are working with two different characteristics from each parent.

It is necessary to use Mendel's law of independent assortment when considering double-factor problems. Recall that according to this law, members of one allelic pair separate from each other independently of the members of other pairs of alleles. This happens during meiosis when the chromosomes segregate. (Mendel's law of independent assortment applies only if the two pairs of alleles are located on separate chromosomes. We will assume this is so in double-factor crosses.)

In humans, the allele for free earlobes is dominant over the allele for attached earlobes. The allele for dark hair dominates the allele for light hair. If both parents are heterozygous for earlobe shape and hair color, what types of offspring can they produce, and what is the probability for each type?

Step 1:

Use the symbol E for free earlobes and e for attached earlobes. Use the symbol D for dark hair and d for light hair.

E = free earlobes D = dark hair
e = attached earlobes d = light hair

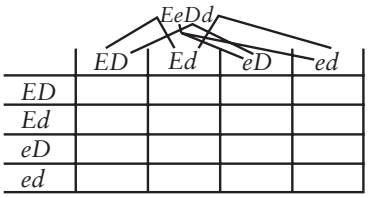
Genotype	*Phenotype*
EE	Free earlobes
Ee	Free earlobes
ee	Attached earlobes
DD	Dark hair
Dd	Dark hair
dd	Light hair

Step 2:
Determine the genotype for each parent and show a mating. The male genotype is *EeDd*, the female genotype is *EeDd*, and the × between them indicates a mating.

EeDd × *EeDd*

Step 3:
Determine all the possible gametes each parent can produce and write the symbols for the alleles in a Punnett square. Because there are two pairs of alleles in a double-factor cross, each gamete must contain one allele from each pair—one from the earlobe pair (either *E* or *e*) and one from the hair color pair (either *D* or *d*). In this example, each parent can produce four different kinds of gametes. The four squares on the left indicate the gametes produced by the male; the four on the top indicate the gametes produced by the female.

To determine the possible gene combinations in the gametes, select one allele from one of the pairs of alleles and match it with one allele from the other pair of alleles. Then match the second allele from the first pair of alleles with each of the alleles from the second pair. This may be done as follows:



Step 4:
Determine all the gene combinations that can result when these gametes unite. Fill in the Punnett square.

	ED	*Ed*	*eD*	*ed*
ED	*EEDD*	*EEDd*	*EeDD*	*EeDd*
Ed	*EEDd*	*EEdd*	*EeDd*	*Eedd*
eD	*EeDD*	*EeDd*	*eeDD*	*eeDd*
ed	*EeDd*	*Eedd*	*eeDd*	*eedd*

Step 5:
Determine the phenotype of each possible gene combination. In this double-factor problem there are 16 possible ways in which gametes can combine to produce offspring. There are four possible phenotypes in this cross. They are represented in the following chart.

Genotype	*Phenotype*	*Symbol*
EEDD or *EEDd* or *EeDD* or *EeDd*	Free earlobes/dark hair	*
EEdd or *Eedd*	Free earlobes/light hair	^
eeDD or *eeDd*	Attached earlobes/dark hair	"
eedd	Attached earlobes/light hair	+

	ED	*Ed*	*eD*	*ed*
ED	*EEDD* *	*EEDd* *	*EeDD* *	*EeDd* *
Ed	*EEDd* *	*Eedd* ^	*EeDd* *	*Eedd* ^
eD	*EeDD* *	*EeDd* *	*eeDD* "	*eeDd* "
ed	*EeDd* *	*Eedd* ^	*eeDd* "	*eedd* +

The probability of having a given phenotype is

9/16 **free earlobes, dark hair**
3/16 **free earlobes, light hair**
3/16 **attached earlobes, dark hair**
1/16 **attached earlobes, light hair**

For our next problem, let's say a man with attached earlobes is heterozygous for hair color and his wife is homozygous for free earlobes and light hair. What can they expect their offspring to be like?

This problem has the same characteristics as the previous problem. Following the same steps, the symbols would be the same, but the parental genotypes would be as follows:

eeDd × *EEdd*

The next step is to determine the possible gametes that each parent could produce and place them in a Punnett square. The male parent can produce two different kinds of gametes, *eD* and *ed*. The female parent can produce only one kind of gamete, *Ed*.

	Ed
eD	
ed	

If you combine the gametes, only two kinds of offspring can be produced:

	Ed
eD	*EeDd*
ed	*Eedd*

They should expect either a child with free earlobes and dark hair or a child with free earlobes and light hair.

10.7 Alternative Inheritance Situations

So far we have considered a few straightforward cases in which a characteristic is determined by simple dominance and recessiveness between two alleles. Other situations, however, may not fit these patterns. Some genetic characteristics are determined by more than two alleles; moreover, some traits are influenced by gene interactions and some traits are inherited differently, depending on the sex of the offspring.

Multiple Alleles and Genetic Heterogeneity

So far we have discussed only traits that are determined by two alleles, for example, *A, a*. However, there can be more

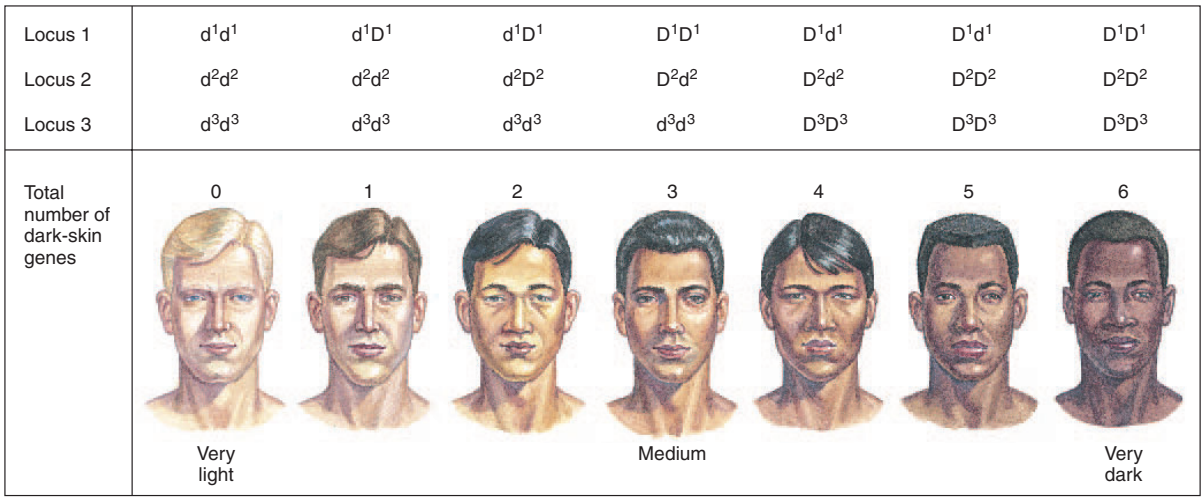


Figure 10.7

Polygenic Inheritance
Skin color in humans is an example of polygenic inheritance. The darkness of the skin is determined by the number of dark-skin genes a person inherits from his or her parents.

than two different alleles for a single trait. All the various forms of the same gene (alleles) that control a particular trait are referred to as **multiple alleles.** However, one person can have only a maximum of two of the alleles for the characteristic. A good example of a characteristic that is determined by multiple alleles is the ABO blood type. There are three alleles for blood type:

Allele*
I^A = blood has type A antigens on red blood cell surface
I^B = blood has type B antigens on red blood cell surface
i **= blood type O has neither type A nor type B antigens on surface of red blood cell**

In the ABO system, A and B show *codominance* when they are together in the same individual, but both are dominant over the O allele. These three alleles can be combined as pairs in six different ways, resulting in four different phenotypes:

Genotype	*Phenotype*
I^AI^A	Blood type A
I^Ai	Blood type A
I^BI^B	Blood type B
I^Bi	Blood type B
I^AI^B	Blood type AB
ii	Blood type O

Multiple-allele problems are worked as single-factor problems.

*The symbols, *I* and *i,* stand for the technical term for the antigenic carbohydrates attached to red blood cells, the *i*mmunogens. These alleles are located on human chromosome 9. The ABO system is not the only one used to type blood. Others include the Rh, MNS, and Xg systems.

Polygenic Inheritance

Thus far we have considered phenotypic characteristics that are determined by alleles at a specific, single place on homologous chromosomes. However, some characteristics are determined by the interaction of genes at several different loci (on different chromosomes or at different places on a single chromosome). This is called **polygenic inheritance.** The fact that a phenotypic characteristic can be determined by many different alleles for a particular characteristic is referred to as **genetic heterogeneity.** A number of different pairs of alleles may combine their efforts to determine a characteristic. Skin color in humans is a good example of this inheritance pattern. According to some experts, genes for skin color are located at a minimum of three loci. At each of these loci, the allele for dark skin is dominant over the allele for light skin. Therefore a wide variety of skin colors is possible depending on how many dark-skin alleles are present (figure 10.7).

Polygenic inheritance is very common in determining characteristics that are quantitative in nature. In the skin-color example, and in many others as well, the characteristics cannot be categorized in terms of *either/or,* but the variation in phenotypes can be classified as *how much* or *what amount* (Outlooks 10.1). For instance, people show great variations in height. There are not just tall and short people—there is a wide range. Some people are as short as 1 meter, and others are taller than 2 meters. This quantitative trait is probably determined by a number of different genes. Intelligence also varies significantly, from those who are severely retarded to those who are geniuses. Many of these traits may be influenced by outside environmental factors such as diet, disease, accidents, and social factors. These are just a few examples of polygenic inheritance patterns.

OUTLOOKS 10.1

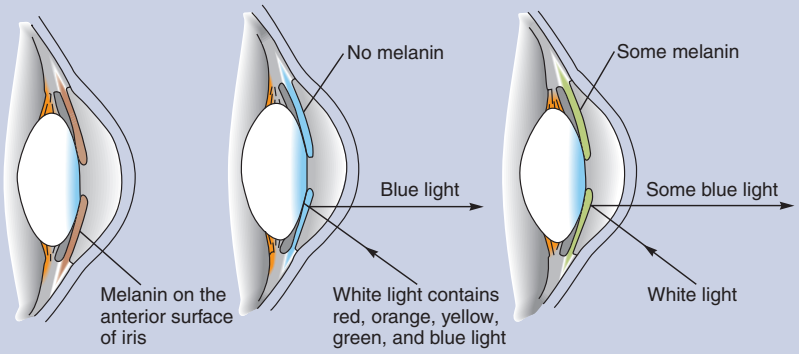
The Inheritance of Eye Color

It is commonly thought that eye color is inherited in a simple dominant/recessive manner. Brown eyes are considered dominant over blue eyes. The real pattern of inheritance, however, is considerably more complicated than this. Eye color is determined by the amount of a brown pigment, known as melanin, present in the iris of the eye. If there is a large quantity of melanin present on the anterior surface of the iris, the eyes are dark. Black eyes have a greater quantity of melanin than brown eyes.

If a large amount of melanin is not present on the anterior surface of the iris, the eyes will appear blue, not because of a blue pigment but because blue light is returned from the iris (see illustration). The iris appears blue for the same reason that deep bodies of water tend to appear blue. There is no blue pigment in the water, but blue wavelengths of light are returned to the eye from the water. People appear to have blue eyes because the blue wavelengths of light are reflected from the iris.

Just as black and brown eyes are determined by the amount of pigment present, colors such as green, gray, and hazel are produced by the various amounts of melanin in the iris. If a very small amount of brown melanin is present in the iris, the eye tends to appear green, whereas relatively large amounts of melanin produce hazel eyes.

Several different genes are probably involved in determining the quantity and placement of the melanin and, therefore, in determining eye color. These genes interact in such a way that a wide range of eye color is possible. Eye color is probably determined by polygenic inheritance, just as skin color and height are. Some newborn babies have blue eyes that later become brown. This is because they have not yet begun to produce melanin in their irises at the time of birth.



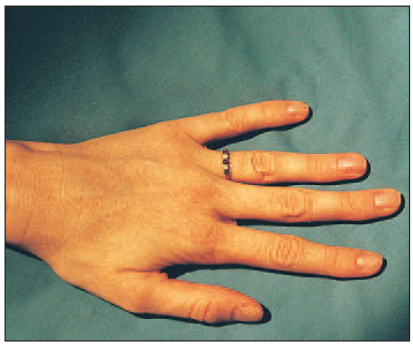
Pleiotropy

Even though a single gene produces only one type of mRNA during transcription, it often has a variety of effects on the phenotype of the person. This is called *pleiotropy*. **Pleiotropy** (*pleio* = changeable) is a term used to describe the multiple effects that a gene may have on the phenotype. A good example of pleiotropy has already been discussed, that is, PKU. In PKU a single gene affects many different chemical reactions that depend on the way a cell metabolizes the amino acid phenylalanine commonly found in many foods (refer to figure 10.6). Another example is *Marfan syndrome* (figure 10.8), a disease suspected to have occurred in former U.S. president, Abraham Lincoln. Marfan syndrome is a disorder of the body's connective tissue but can also have effects in many other organs including the eyes, heart, blood, skeleton, and lungs. Symptoms generally appear as a tall, lanky body with long arms and spider fingers, scoliosis, osteoporosis, and depression or protrusion of the chest wall (funnel chest/pectus excavatum or pigeon chest/pectus carinatum). In many cases these nearsighted people also show dislocation of the lens of the eye. The white of the eye (sclera) may appear bluish. Heart problems include dilation of the aorta and prolapse of the heart's mitral valve. Death may be caused by a dissection (tear) in the aorta from the rupture in a weakened and dilated area of the aorta, called an aortic aneurysm.

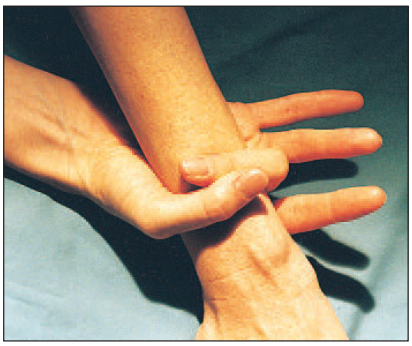
10.8 Environmental Influences on Gene Expression

Maybe you assumed that the dominant allele would always be expressed in a heterozygous individual. It is not so simple! Here, as in other areas of biology, there are exceptions. For example, the allele for six fingers (*polydactylism*) is dominant over the allele for five fingers in humans. Some people who have received the allele for six fingers have a fairly complete sixth finger; in others, it may appear as a little stub. In another case, a dominant allele causes the formation of a little finger that cannot be bent like a normal little finger. However, not all people who are believed to have inherited that allele will have a stiff little finger. In some cases, this dominant characteristic is not expressed or perhaps only shows on one hand. Thus, there may be variation in the

(a)



(b)



(c)

Figure 10.8

Marfan Syndrome
It is estimated that about 40,000 (the incidence is 1 out of 10,000) people in the United States have this autosomal dominant abnormality. Notice the lanky appearance to the body and face of this person with Marfan syndrome (*a*). Photos (*b*) and (*c*) illustrate their unusually long fingers.

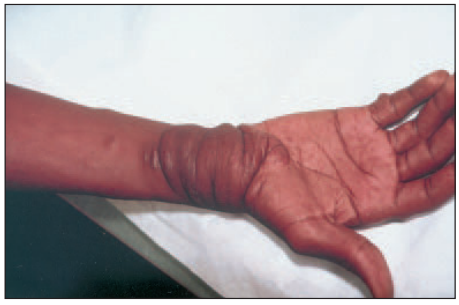


Figure 10.9

Neurofibromatosis 1
This abnormality is seen in many forms including benign fibromatous skin tumors, "café au lait" spots, nodules in the iris, and possible malignant tumors. It is extremely variable in its expressivity, i.e., the traits may be almost unnoticeable or extensive. An autosomal dominant trait, it is the result of a mutation and the production of a protein (neurofibromin) that normally would suppress the activity of a gene that causes tumor formation.

Figure 10.10

Baldness and the Expression of Genes
It is a common misconception that males have genes for baldness and females do not. Male-pattern baldness is a sex-influenced trait in which both males and females may possess alleles coding for baldness. These genes are turned on by high levels of the hormone testosterone. This is another example of an internal gene-regulating factor.

degree to which an allele expresses itself *in an individual*. Geneticists refer to this as *variable expressivity*. A good example of this occurs in the genetic abnormality *neurofibromatosis type 1 (NF1)* (figure 10.9). In some cases it may not be expressed *in the population* at all. This is referred to as a *lack of penetrance*. Other genes may be interacting with these dominant alleles, causing the variation in expression.

Both internal and external environmental factors can influence the expression of genes. For example, at conception, a male receives genes that will eventually determine the pitch of his voice. However, these genes are expressed differently after puberty. At puberty, male sex hormones are released. This internal environmental change results in the deeper male voice. A male who does not produce these hormones retains a higher-pitched voice in later life. Another characteristic whose expression is influenced by internal gene-regulating mechanisms is that of male-pattern baldness (figure 10.10).

A comparable situation in females occurs when an abnormally functioning adrenal gland causes the release of large amounts of male hormones. This results in a female with a deeper voice. Also recall the genetic disease PKU. If

children with *phenylketonuria (PKU)* are allowed to eat foods containing the amino acid phenylalanine, they will become mentally retarded. However, if the amino acid phenylalanine is excluded from the diet, and certain other dietary adjustments are made, the person will develop normally. NutraSweet is a phenylalanine-based sweetener, so people with this genetic disorder must use caution when buying products that contain it.

Diet is an external environmental factor that can influence the phenotype of an individual. *Diabetes mellitus*, a metabolic disorder in which glucose is not properly metabolized and is passed out of the body in the urine, has a genetic basis. Some people who have a family history of diabetes are thought to have inherited the trait for this disease. Evidence indicates that they can delay the onset of the disease by reducing the amount of sugar in their diet. This change in the external environment influences gene expression in much the same way that sunlight affects the expression of freckles in humans (see figure 10.3).

SUMMARY

Genes are units of heredity composed of specific lengths of DNA that determine the characteristics an organism displays. Specific genes are at specific loci on specific chromosomes. The phenotype displayed by an organism is the result of the effect of the environment on the ability of the genes to express themselves.

Diploid organisms have two genes for each characteristic. The alternative forms of genes for a characteristic are called alleles. There may be many different alleles for a particular characteristic. Organisms with two identical alleles are homozygous for a characteristic; those with different alleles are heterozygous. Some alleles are dominant over other alleles that are said to be recessive.

Sometimes two alleles express themselves, and often a gene has more than one recognizable effect on the phenotype of the organism. Some characteristics may be determined by several different pairs of alleles. In humans and some other animals, males have an X chromosome with a normal number of genes and a Y chromosome with fewer genes. Although they are not identical, they behave as a pair of homologous chromosomes. Because the Y chromosome is shorter than the X chromosome and has fewer genes, many of the recessive characteristics present on the X chromosome appear more frequently in males than in females, who have two X chromosomes.

CONCEPT MAP TERMINOLOGY

Construct a concept map to show relationships among the following concepts.

law of independent assortment
locus
offspring
probability
recessive allele
single-factor inheritance
X-linked trait

KEY TERMS

alleles
autosomes
carrier
codominance
dominant allele
double-factor cross
gene
gene expression
genetic heterogeneity
genetics
genome
genotype
heterozygous
homozygous
law of dominance
law of independent assortment
law of segregation
linkage group
locus (loci)
Mendelian genetics
multiple alleles
offspring
phenotype
pleiotropy
polygenic inheritance
probability
Punnett square
recessive allele
sex chromosomes
single-factor cross
X-linked gene

e—LEARNING CONNECTIONS *www.mhhe.com/enger10*

Topics	Questions	Media Resources
10.1 Genetics, Meiosis, and Cells	1. How many kinds of gametes are possible with each of the following genotypes? a. *Aa* b. *AaBB* c. *AaBb* d. *AaBbCc*	**Quick Overview** • Mathematical description of meiosis **Key Points** • Genetics, meiosis, and cells
10.2 Single-Gene Inheritance Patterns		**Quick Overview** • Simple types of allele interactions **Key Points** • Single-gene inheritance patterns

Topics	Questions	Media Resources
10.3 Mendel's Laws of Heredity		**Quick Overview** • Rules of thumb for genetics problems **Key Points** • Mendel's laws of heredity
10.4 Probability Versus Possibility		**Quick Overview** • Brushing up your math skills **Key Points** • Probability versus possibility
10.5 Steps in Solving Heredity Problems: Single-Factor Crosses		**Quick Overview** • Learning the strategy to story problems **Key Points** • Steps in solving heredity problems: Single-factor crosses
10.6 The Double-Factor Cross	2. What is the probability of each of the following sets of parents producing the given genotypes in their offspring? **Parents** — **Offspring** a. *AA* × *aa* — *Aa* b. *Aa* × *Aa* — *Aa* c. *Aa* × *Aa* — *aa* d. *AaBb* × *AaBB* — *AABB* e. *AaBb* × *AaBB* — *AaBb* f. *AaBb* × *AaBb* — *AABB* 3. If an offspring has the genotype *Aa,* what possible combinations of parental genotypes exist?	**Quick Overview** • Expanding your strategy **Key Points** • The double-factor cross **Animations and Review** • Dihybrid cross **Interactive Concept Maps** • Text concept map
10.7 Alternative Inheritance Situations	4. In humans, the allele for albinism is recessive to the allele for normal skin pigmentation. a. What is the probability that a child of a mother and a father who are heterozygous will be albino? b. If a child is normal, what is the probability that it is a carrier of the albino allele? 5. In certain pea plants, the allele *T* for tallness is dominant over *t* for shortness. a. If a homozygous tall and homozygous short plant are crossed, what will be the phenotype and genotype of the offspring? b. If both individuals are heterozygous, what will be the phenotypic and genotypic ratios of the offspring? 6. What is the probability of a child having type AB blood if one of the parents is heterozygous for A blood and the other is heterozygous for B? What other genotypes are possible in this child?	**Quick Overview** • New ways to understand allele interactions **Key Points** • Alternative inheritance situations **Animations and Review** • Beyond Mendel **Experience This!** • Chart your own pedigree **Case Study** • Should you need a license to be a parent?
10.8 Environmental Influences on Gene Expression		**Quick Overview** • Nature versus nurture? **Key Points** • Environmental influences on gene expression