7.1 Chromosomes and Phenotype

KEY CONCEPT The chromosomes on which genes are located can affect the expression of traits.

MAIN IDEAS

- Two copies of each autosomal gene affect phenotype.
- Males and females can differ in sex-linked traits.

VOCABULARY

carrier, p. 201
sex-linked gene, p. 201
X chromosome inactivation, p. 203

Review
dominant, recessive, phenotype, allele, gene, autosome, sex chromosome, trait

REVIEW AT CLASSZONE.COM

Connect The next time you are in a crowd of people, take a moment to look at the variety of traits around you. Hair color and texture, eye color and shape, height, and weight are all influenced by genetics. Can dominant and recessive alleles of one gene produce so many subtle differences in any of those traits? In most cases, the answer is no. But the dominant and recessive relationship among alleles is a good place to start when learning about the complexities of genetics.

MAIN IDEA

Two copies of each autosomal gene affect phenotype.

You read in Chapter 6 how some genetic traits depend on dominant and recessive alleles. But many factors affect phenotype, including the specific chromosome upon which a gene is located. Gene expression is often related to whether a gene is located on an autosome or on a sex chromosome. Recall that sex chromosomes determine an organism's sex. Autosomes are all of the other chromosomes, and they do not play a direct role in sex determination.

You also know that sexually reproducing organisms have two of each chromosome. Each pair consists of one chromosome from each of two parents. Both chromosomes have the same genes, but the chromosomes might have different alleles for those genes. And, as Mendel observed, different alleles can produce different phenotypes, such as white flowers or purple flowers.

All of the traits that Mendel studied are determined by genes on autosomes. In fact, most traits in sexually reproducing organisms, including humans, are the result of autosomal genes. Look at **FIGURE 7.1.** Is your hair curly or straight? What about your parents' hair? The genes that affect your hair texture—curly hair or straight hair—are autosomal genes.

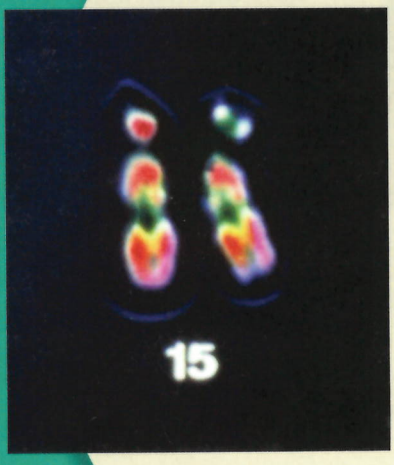
Many human genetic disorders are also caused by autosomal genes. The chance of a person having one of these disorders can be predicted, just as Mendel could predict the phenotypes that would appear in his pea plants. Why? Because there are two copies of each gene on autosomes—one on each homologous chromosome—and each copy can influence phenotype.

FIGURE 7.1 Hair texture is just one example of a trait that is controlled by autosomal genes.

Why are there so many variations among people?

Connecting CONCEPTS

It will come as no surprise to you, but you are not a pea plant. But, Mendel's principles apply to you just as they apply to other organisms. About 99.9 percent of everyone's DNA is identical. So how can a 0.1 percent difference in DNA lead to the wide range of human traits? In many organisms, genetics is more than dominant and recessive alleles.



Multiple Gene Traits Two genes for human eye color are located on chromosome 15, shown at the left. One reason for the large variations in phenotype in many species is that most traits are produced by several genes that interact with each other. Eye color in humans is a trait controlled by more than one gene. And the alleles of those genes have different dominant and recessive relationships. (colored LM; magnification 13,000×)

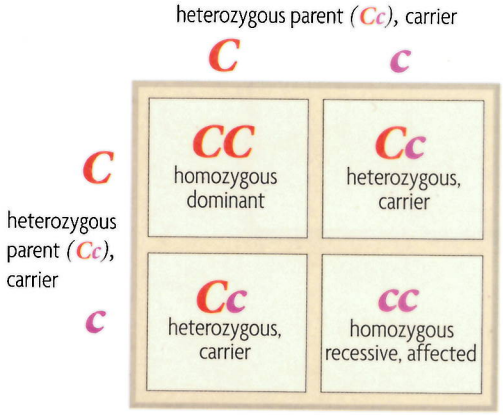
Disorders Caused by Recessive Alleles

Some human genetic disorders are caused by recessive alleles on autosomes. Two copies of the recessive allele must be present for a person to have the disorder. These disorders often appear in offspring of parents who are both heterozygotes. That is, each parent has one dominant, "normal" allele that masks the one disease-causing recessive allele.

For example, cystic fibrosis is a severe recessive disorder that mainly affects the sweat glands and the mucus glands. A person who is homozygous for the recessive allele will have the disease. Someone who is heterozygous for the alleles will not have the disease, but is a carrier. A **carrier** does not show disease symptoms, but can pass on the disease-causing allele to offspring. In this way, alleles that are lethal, or deadly, in a homozygous recessive individual can remain in a population's gene pool. This inheritance pattern is shown in **FIGURE 7.2.**

FIGURE 7.2 AUTOSOME INHERITANCE

Some genetic disorders, such as cystic fibrosis, are inherited according to Mendel's principles.



Disorders Caused by Dominant Alleles

Dominant genetic disorders are far less common than recessive disorders. One example is Huntington's disease. Huntington's disease damages the nervous system and usually appears during adulthood. Because the disease is caused by a dominant allele, there is a 50 percent chance that a child will have it even if only one parent has one of the alleles. If both parents are heterozygous for the disease, there is a 75 percent chance that any of their children will inherit the disease. Because Huntington's disease strikes later in life, a person with the allele can have children before the disease appears. In that way, the allele is passed on in the population even though the disease is fatal.

Connect **How are Mendel's observations related to genes on autosomes?**

MAIN IDEA

Males and females can differ in sex-linked traits.

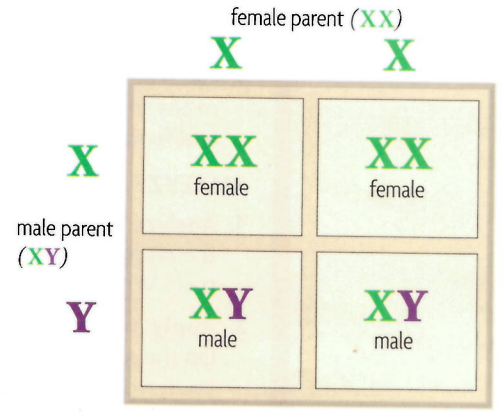
Mendel figured out much about heredity, but he did not know about chromosomes. As it turns out, he only studied traits produced by genes on autosomes. Now, we know about sex chromosomes, and we know that the expression of genes on the sex chromosomes differs from the expression of autosomal genes.

Sex-Linked Genes

Genes that are located on the sex chromosomes are called **sex-linked genes.** Recall from Chapter 6 that many species have specialized sex chromosomes called the X and Y chromosomes. In mammals and some other animals, individuals with two X chromosomes—an XX genotype—are female. Individuals with one X and one Y—an XY genotype—are male. As **FIGURE 7.3** shows, a female can pass on only an X chromosome to offspring, but a male can pass on either an X or a Y chromosome.

FIGURE 7.3 SEX CHROMOSOME INHERITANCE

The gametes from an XY male determine the sex of the offspring.



TAKING NOTES

Use a two-column chart to compare and contrast the expression of autosomal and sex-linked genes.

autosomes	sex chromosomes

Genes on the Y chromosome are responsible for the development of male offspring, but the X chromosome actually has much more influence over phenotype. The X chromosome has many genes that affect many traits. Scientists hypothesize that the Y chromosome may have genes for more than sex determination, but there is little evidence to support this idea.

In many organisms, including humans, the Y chromosome is much smaller and has many fewer genes than the X chromosome. Evidence suggests that over millions of years of evolution, the joining of the X and Y chromosomes during meiosis has resulted in segments of the Y chromosome being transferred to the X. You will read more about specific sex-linked genes and their locations on the human X and Y chromosomes in Section 7.4.

Expression of Sex-Linked Genes

Because the X and Y chromosomes have different genes, sex-linked genes have a pattern of expression that is different from autosomal genes. Remember, tw copies of an autosomal gene affect a trait. What happens when there is only one copy of a gene, as is the case in an XY male? Because males have only on copy of each type of sex chromosome, they express all of the alleles on both chromosomes. In males, there are no second copies of sex-linked genes to mask the effects of another allele. This means that even if all of the alleles c sex-linked genes in a male are recessive, they will be expressed.

QUICK LAB **PREDICTING**

Sex-Linked Inheritance

The relationship between genotype and phenotype in sex-linked genes differs from that in autosomal genes. A female must have two recessive alleles of a sex-linked gene to express a recessive sex-linked trait. Just one recessive allele is needed for the same trait to be expressed in a male. In this lab, you will model the inheritance pattern of sex-linked genes.

PROBLEM How does probability explain sex-linked inheritance?

MATERIALS

- 2 coins
- masking tape
- marker
- index card with genetic cross

PROCEDURE

1. Use the tape and marker to label two coins with the genetic cross shown on your group's index card. One coin represents the egg cell and the other coin represents the sperm cell.
2. Flip the two coins and record the genotype of the "offspring."
3. Repeat step 2 until you have modeled 50 genetic crosses. Make a data table to record each genetic cross that you model.
4. Calculate the genotype and phenotype probabilities for both males and females. Calculate the frequency of male offspring and female offspring.

ANALYZE AND CONCLUDE

1. **Analyze** Do all of the females from the genetic cross show the recessive trait? Do all of the males show the recessive trait? Why or why not?
2. **Apply** Make a Punnett square that shows the genetic cross. Do the results from your Punnett square agree with those from your experiment? Why or why not?

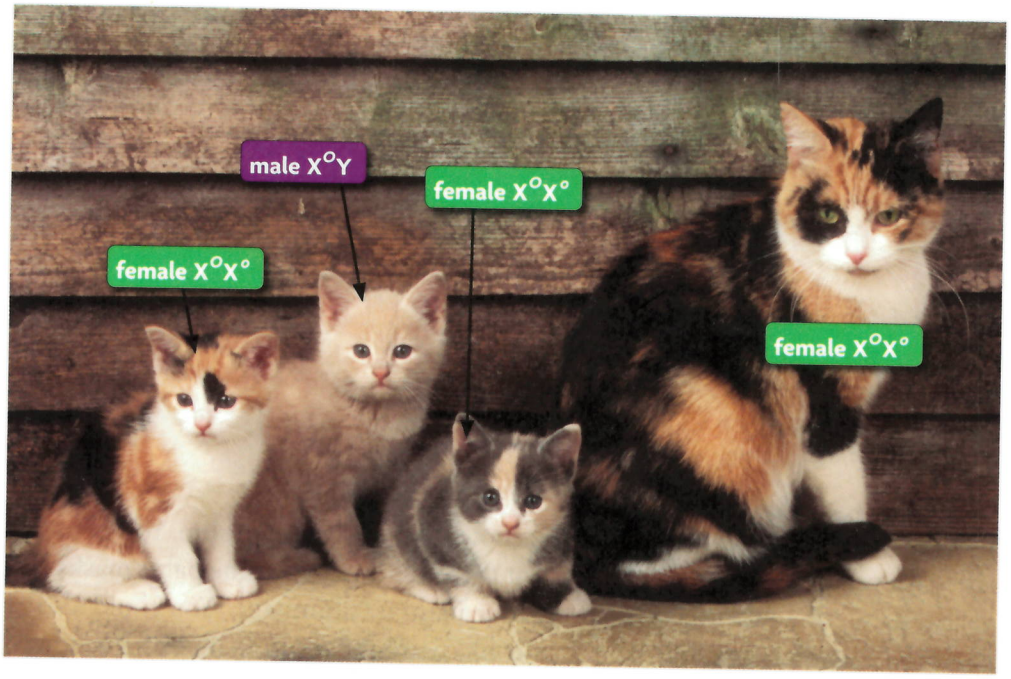


FIGURE 7.4 The female calico cats have two X chromosomes with different alleles for fur color. Both alleles are expressed in a random pattern. The male cat has only one X chromosome, and its allele for fur color is expressed across the entire body.

X^{O} = Orange fur allele
X^{o} = Black fur allele

In mammals, the expression of sex-linked genes in females is also different from the way in which genes on other chromosomes are expressed. In each cell of female mammals, one of the two X chromosomes is randomly "turned off" by a process called **X chromosome inactivation.** Because of X chromosome inactivation, females are a patchwork of two types of cells—one type with an active X chromosome that came from the mother, and a second type with an active X chromosome that came from the father.

Colorful examples of X chromosome inactivation are seen in female tortoiseshell cats and female calico cats. The female calico cats shown in **FIGURE 7.4** have white fur, as well as alleles for black or orange fur on their X chromosomes. Those alleles are expressed randomly in cells across the cat's body. As a result, its coat is a mixture of color splotches. It is truly a patchwork of cells. Because the male cats only have one X chromosome, they have white fur and one sex-linked gene for either orange or black fur.

Infer **Why are males more likely than females to have sex-linked genetic disorders?**

Connecting CONCEPTS

Mitosis Recall from **Chapter 5** that DNA coils to form chromosomes. In XX females, one of the two X chromosomes in each cell is "inactivated" by becoming even more tightly coiled.

7.1 ASSESSMENT

ONLINE QUIZ ClassZone.com

REVIEWING MAIN IDEAS

1. How are autosomal traits, including recessive genetic disorders that are carried in a population, related to Mendel's observations of heredity?
2. Describe how **sex-linked genes** are expressed differently in males and in females.

CRITICAL THINKING

3. **Apply** How might a scientist determine whether a trait is sex-linked by observing the offspring of several genetic crosses?
4. **Compare and Contrast** How is the expression of sex-linked genes both similar to and different from the expression of autosomal genes?

Connecting CONCEPTS

5. **Meiosis** Scientists hypothesize that over millions of years, the Y chromosome has lost genes to the X chromosome. During what stages of meiosis might the Y chromosome have transferred genes to the X chromosome? Explain.

7.2 Complex Patterns of Inheritance

KEY CONCEPT Phenotype is affected by many different factors.

MAIN IDEAS

- Phenotype can depend on interactions of alleles.
- Many genes may interact to produce one trait.
- The environment interacts with genotype.

VOCABULARY

incomplete dominance, p. 204
codominance, p. 205
polygenic trait, p. 206

Review
allele, phenotype, genotype

REVIEW AT CLASSZONE.COM

Connect Suppose you have blue and yellow paints to paint a room. You paint the walls yellow, let them dry, then paint the walls blue. The blue paint masks the yellow paint, so you could say that the blue paint is "dominant." You could also combine the paints in other ways. You could paint the room in blue and yellow stripes, or you could mix the colors and paint the room green. You can think of different alleles as different paint colors, but in genetics there are many more paint colors—alleles—and many more ways that they are combined.

MAIN IDEA

Phenotype can depend on interactions of alleles.

Although Mendel's basic theory of heredity was correct, his research could not have explained all of the continuous variations for many traits. For example, many traits result from alleles with a range of dominance, rather than a strict dominant and recessive relationship.

Connecting CONCEPTS

Principles of Genetics Recall from **Chapter 6** that a homozygote has two identical alleles of a gene, and a heterozygote has two different alleles of a gene.

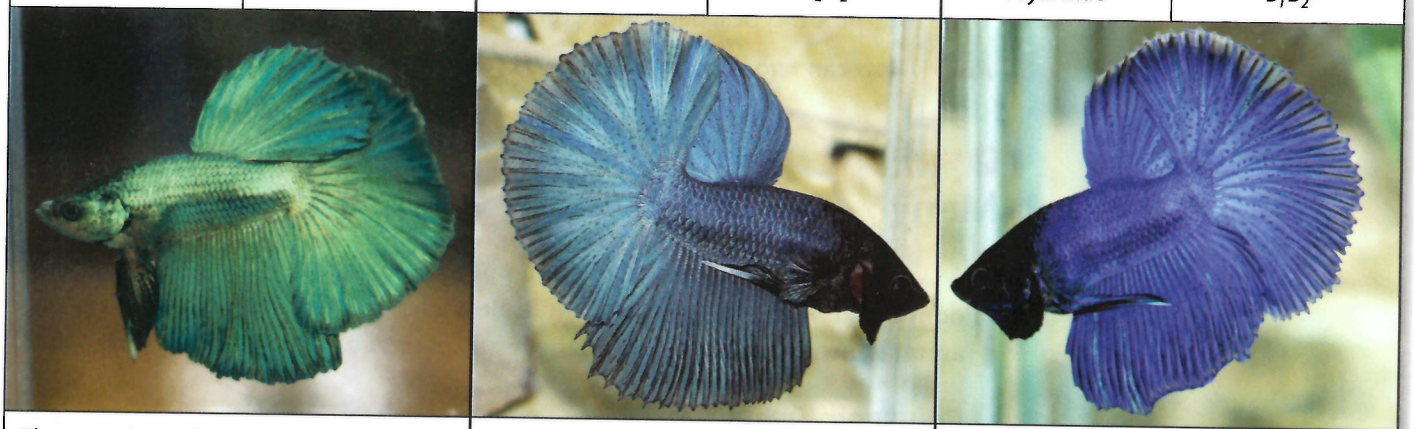
The pea flowers that Mendel observed were either white or purple. One allele was dominant, but dominance does not mean that one allele "defeats" the other. Usually, it means that the dominant allele codes for a certain protein and the recessive allele codes for a variation of the protein that has little or no effect. In Mendel's pea flowers, a heterozygous plant makes enough of the purple color that only one dominant allele is needed to give the flowers a purple color. But in many cases, a phenotype comes from more than just one gene, and many genes in a population have more than just two alleles.

Incomplete Dominance

Sometimes, alleles show **incomplete dominance,** in which a heterozygous phenotype is somewhere between the two homozygous phenotypes. Neither allele is completely dominant nor completely recessive. One example of incomplete dominance is the four-o'clock plant. When plants that are homozygous for red flowers are crossed with plants that are homozygous for white flowers, the offspring have pink flowers. The pink color is a third, distinct phenotype. Neither of the original phenotypes of the plants in the parent's generation can be seen separately in the F_1 generation offspring.

FIGURE 7.5 Incomplete Dominance

PHENOTYPE	GENOTYPE	PHENOTYPE	GENOTYPE	PHENOTYPE	GENOTYPE
green	B_1B_1	steel blue	B_2B_2	royal blue	B_1B_2



The green betta fish is homozygous for the green color allele.

The steel blue betta fish is homozygous for the blue color allele.

The royal blue betta fish is heterozygous for the two color alleles.

Another example of incomplete dominance is the color of betta fish shown in **FIGURE 7.5.** When a green fish (B_1B_1) is crossed with a steel blue fish (B_2B_2), all of the offspring have the heterozygous genotype (B_1B_2). These offspring will be a royal blue color that comes from the phenotypes from both alleles. The alleles of this gene follow a pattern of incomplete dominance. What happens when two royal blue betta fish are crossed? Some offspring (25 percent) will be green (B_1B_1), some (50 percent) will be royal blue (B_1B_2), and some (25 percent) will be steel blue (B_2B_2).

VOCABULARY

When alleles are neither dominant nor recessive, such as with incomplete dominance, uppercase letters with either subscripts or superscripts are used to represent the different alleles.

Codominance

Sometimes, both alleles of a gene are expressed completely—neither allele is dominant nor recessive. In this case, alleles show **codominance,** in which both traits are fully and separately expressed. Suppose a plant that is homozygous for red flowers is crossed with a plant that is homozygous for white flowers. In incomplete dominance, the offspring have pink flowers. Codominant alleles are different. Instead of what looks like an intermediate phenotype, both traits are expressed. The flowers will have some red areas and some white areas.

One trait that you likely know about—human ABO blood types—is an example of codominance. And, because the blood types come from three different alleles in the human population, this trait is also considered a multiple-allele trait. The multiple alleles, shown in **FIGURE 7.6,** are called I^A, I^B, and i. Both I^A and I^B result in a protein, called an antigen, on the surface of red blood cells. Allele i is recessive and does not result in an antigen. Someone with a genotype of I^Ai will have type A blood, and someone with a genotype of I^Bi will have type B blood. But remember that the I^A and I^B alleles are codominant.

FIGURE 7.6 CODOMINANCE

PHENOTYPE (BLOOD TYPE)		GENOTYPES
A	antigen A	I^AI^A or I^Ai
B	antigen B	I^BI^B or I^Bi
AB	both antigens	I^AI^B
O	no antigens	ii

People with both codominant alleles (I^AI^B) have both antigens, so they have type AB blood. People with an *ii* genotype have red blood cells without either antigen, and they have type O blood. Two heterozygous people, one with type A blood (I^Ai) and one with type B blood (I^Bi), can have offspring with any of the four blood types, depending on the alleles that are passed on.

Apply **How can two people with type B blood have a child with type O blood?**

MAIN IDEA

Many genes may interact to produce one trait.

As you have seen, some variations in phenotype are related to incomplete dominance, codominance, and multiple alleles. But most traits in plants and animals, including humans, are the result of several genes that interact.

Polygenic Traits

Traits produced by two or more genes are called **polygenic traits.** Human skin color, for example, is the result of four genes that interact to produce a continuous range of colors. Similarly, human eye color, which is often thought of as a single gene trait, is polygenic. As **FIGURE 7.7** shows, at least three genes with complicated patterns of expression play roles in determining eye color. For example, the green allele is dominant to blue alleles, but it is recessive to all brown alleles. These genes do not account for all eye color variations, such as changes in eye color over time, the continuous range of eye colors, and patterns of colors in eyes. As a result, scientists hypothesize that still undiscovered genes affect eye color.

VISUAL VOCAB

Traits that are produced by two or more genes are called **polygenic traits.**

many + genes → poly genic

Epistasis

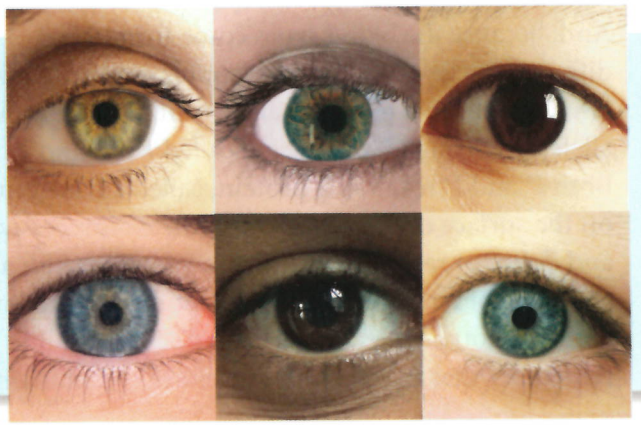
Another polygenic trait is fur color in mice and in other mammals. In mice, at least five different genes interact to produce the phenotype. Two genes give the mouse its general color, one gene affects the shading of the color, and one gene determines whether the mouse will have spots. But the fifth gene involved in mouse fur color can overshadow all of the others. In cases such as this, one gene, called an epistatic gene, can interfere with the expression of other genes.

FIGURE 7.7 Eye Color

At least three different genes interact to produce the range of human eye colors, such as in the examples on the right.

GENE NAME	DOMINANT ALLELE	RECESSIVE ALLELE
BEY1	brown	blue
BEY2	brown	blue
GEY	green	blue

Order of dominance: brown > green > blue.



In albinism, a single epistatic gene interferes with the expression of other genes. Albinism, as you can see in **FIGURE 7.8**, is characterized by a lack of pigment in skin, hair, and eyes. A mouse that is homozygous for the alleles that prevent the coloration of fur will be white, regardless of the phenotypes that would normally come from the other four genes. A person with the alleles for albinism will have very light skin, hair, and eyes, regardless of the other genes he or she has inherited.

Contrast **How do multiple-allele traits differ from polygenic traits?**

FIGURE 7.8 Albinism in mammals, such as this hedgehog, is caused by an epistatic gene that blocks the production of pigments.

MAIN IDEA

The environment interacts with genotype.

Phenotype is more than the sum of gene expression. For example, the sex of sea turtles depends both on genes and on environment. Female turtles make nests on beaches and bury their eggs in the sand. Eggs that mature in warmer temperatures develop into female turtles. Eggs that mature in cooler temperatures develop into male turtles.

Genes and environment also interact to determine human traits. Think about height. Genes give someone a tendency to be either short or tall, but they do not control everything. An interesting question for the interaction between genes and environment is "Are identical twins always identical?" Studies of identical twins have shown that the environment during early development can have long-lasting effects. One twin might get more nutrients than the other because of its position in the mother's uterus. This difference can result in height and size differences that last throughout the twins' lives. Also, twins raised in environments with different nutrition and health care often differ in height and other physical traits. In the end, phenotype is usually a mixture of genes and environment.

Connect **Sunlight can cause a person's hair to become lighter in color. Is this an example of an interaction between genes and the environment? Why or why not?**

NSTA scilinks.org SCILINKS

Find out more about dominant and recessive traits at scilinks.org.
Keycode: MLB007

7.2 ASSESSMENT

ONLINE QUIZ ClassZone.com

REVIEWING MAIN IDEAS

1. How is **incomplete dominance** expressed in a phenotype?
2. Why might **polygenic traits** vary more in phenotype than do single-gene traits?
3. Explain how interactions between genes and the environment can affect phenotype.

CRITICAL THINKING

4. **Synthesize** How is **codominance** the same as having no dominant and recessive relationship at all between two alleles?
5. **Compare and Contrast** How are codominant alleles and incompletely dominant alleles similar? How are they different?

Connecting CONCEPTS

6. **Principles of Genetics** Why can parents who are heterozygous for type A and type B blood have children with any of the four human blood types? Use a Punnett square to support your answer.

Codominance

Codominant alleles are both expressed in a person's phenotype. A heterozygote will have the traits associated with both alleles. In this lab, you will explore codominance by analyzing the results of tests for sickle cell disease within a family.

MATERIALS

- paper
- pencil

PROCESS SKILLS

- Inferring
- Predicting

BACKGROUND

Sickle cell disease is caused by a change in the gene for hemoglobin, which is the oxygen-carrying protein in red blood cells. Individuals who are homozygous for the sickle cell allele often cannot endure exercise. Individuals who are heterozygous for the allele can have sickle cell attacks under extreme conditions. Normal individuals ($Hb^S\ Hb^S$) have only normal hemoglobin. Homozygous sickle cell individuals ($Hb^s\ Hb^s$) have only sickle cell hemoglobin. Heterozygous individuals ($Hb^S\ Hb^s$) have both normal hemoglobin and sickle cell hemoglobin.

Jerry Smith collapsed while running a race for his track team. A doctor said that he had a sickle cell attack. Genetic tests were run on several family members. The test results are shown below. An X indicates that form of hemoglobin in red blood cells.

PROBLEM How can you determine the genotypes of people in a family?

TABLE 1. FAMILY PHENOTYPES

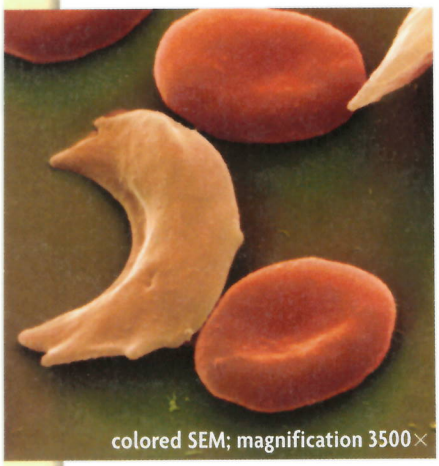
Subject	Normal Hemoglobin	Sickle Cell Hemoglobin
Jerry Smith	X	X
Jerry's brother	X	
Jerry's younger sister	X	X
Jerry's youngest sister	X	
Jerry's father	X	
Jerry's grandfather	X	
Jerry's grandmother	X	X

PROCEDURE

1. Use the background information and the genetic test results to answer questions 1–4.
2. Use the background information and a Punnett square to help you answer question 5.

ANALYZE AND CONCLUDE

1. **Analyze** Are any of Jerry's siblings homozygous for the sickle cell allele? Are any of Jerry's siblings heterozygous for sickle cell disease?
2. **Analyze** What genotype is Jerry's father?
3. **Analyze** What genotypes are Jerry's grandparents?
4. **Infer** What is the genotype of Jerry's mother? Explain.
5. **Predict** If Jerry marries a female who is heterozygous for the sickle cell allele, what would be the possible genotypes and phenotypes of their children, according to your Punnett square?



colored SEM; magnification 3500×

7.3 Gene Linkage and Mapping

KEY CONCEPT Genes can be mapped to specific locations on chromosomes.

MAIN IDEAS

- Gene linkage was explained through fruit flies.
- Linkage maps estimate distances between genes.

VOCABULARY

linkage map, p. 210

Review

chromosome, crossing over

REVIEW AT CLASSZONE.COM

Connect If you leave a banana out on a table until it is very ripe, you might see some of the most useful organisms for genetic research—fruit flies—buzzing around it. In your kitchen, fruit flies are pests. In the laboratory, early experiments with fruit flies showed not only that genes are on chromosomes but also that genes are found at specific places on chromosomes.

MAIN IDEA

Gene linkage was explained through fruit flies.

Gene linkage, which you read about in Chapter 6, was first described by William Bateson and R. C. Punnett, who invented the Punnett square. Punnett and Bateson, like Mendel, studied dihybrid crosses of pea plants. But their results differed from the 9:3:3:1 phenotype ratios that Mendel observed. The results suggested that some genes were linked together. But how could genes be linked and still follow Mendel's law of independent assortment?

American scientist Thomas Hunt Morgan, who worked with fruit flies (*Drosophila melanogaster*), found the answer. At first, Morgan was just looking for an organism to use in genetic research. He found fruit flies very useful because he could quickly and cheaply grow new generations of flies. He observed among fruit flies easily identifiable variations in eye color, body color, and wing shape. Knowing these variations, Morgan and his students set up experiments similar to Mendel's dihybrid crosses. They chose one type of fly with traits associated with the wild type, or most common phenotype. They crossed the wild type flies with mutant flies, or flies with a different, less common phenotype. You can see examples of fruit flies in **FIGURE 7.9.**

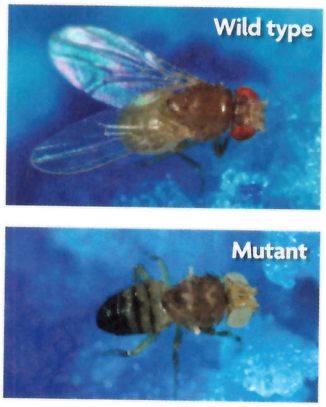


FIGURE 7.9 The wild type fruit fly (top) shows the most common phenotype. The mutant fruit fly (bottom) has no wings, white eyes, and a different body color.

Morgan's results, like those of Punnett and Bateson, did not always follow the 9:3:3:1 ratio predicted by Mendel. But the results did differ in a noticeable pattern. Some traits appeared to be inherited together. Morgan called these traits linked traits, and they appeared to fall into four groups. As it turns out, fruit flies have four pairs of chromosomes. Each of the four groups of linked traits identified by Morgan matches one of the chromosome pairs. Morgan concluded that linked genes were on the same chromosome. The chromosomes, not the genes, assort independently during meiosis. Because the linked genes were not inherited together every time, Morgan also concluded that chromosomes must exchange homologous genes during meiosis.

Synthesize **How did Morgan's research build upon Mendel's observations?**

DATA ANALYSIS

CONSTRUCTING BAR GRAPHS

Scientists tested the reaction of fruit flies to stress by exposing them to bright light—a source of stress for *Drosophila*. The scientists timed how long it took for half of the flies in each group to reach food, which they called a "half-time." Three strains of flies were tested—wild type 1, wild type 2, and a mutant eyeless type—under control conditions and with bright light. The data are shown in Table 1.

1. **Graph Data** Construct a bar graph that shows the data in the table. Recall that the independent variable is on the *x*-axis and the dependent variable is on the *y*-axis.
2. **Analyze** How did the condition of bright light affect the flies? Were all strains affected to the same degree? Why or why not?

TABLE 1. *DROSOPHILA* RESPONSES TO LIGHT

Strain	Condition	Half-Time (min)
Wild type 1	control	4.0
Wild type 1	bright light	12.5
Wild type 2	control	4.5
Wild type 2	bright light	12.0
Eyeless	control	4.5
Eyeless	bright light	5.0

Source: V. Min, B. Condron, Journal of Neuroscience Methods, 145.

MAIN IDEA

Linkage maps estimate distances between genes.

The probability that two genes on a chromosome will be inherited together is related to the distance between them. The closer together two genes are, the more likely it is that they will be inherited together. The farther apart two genes are, the more likely it is that they will be separated during meiosis.

One of Morgan's students, Alfred Sturtevant, hypothesized that the frequency of cross-overs during meiosis was related to the distance between genes. This meant that the closer together two genes were, the more likely they were to stay together when cross-overs took place. Sturtevant identified three linked traits in fruit flies—body color, eye color, and wing size—and then crossed the fruit flies. He recorded the percentage of times that the phenotypes did not appear together in the offspring. This percentage represented the frequency of cross-overs between chromosomes.

From the cross-over frequencies, Sturtevant made **linkage maps,** which are maps of the relative locations, or loci, of genes on a chromosome. On a linkage map, one map unit is equal to one cross-over for each 100 offspring, or one percentage point. You can see an example of a linkage map in **FIGURE 7.10.**

Making a linkage map is fairly easy if all of the cross-over frequencies for the genes being studied are known. Suppose the following data were collected.

- Gene A and gene B cross over 6.0 percent of the time.
- Gene B and gene C cross over 12.5 percent of the time.
- Gene A and gene C cross over 18.5 percent of the time.

According to Sturtevant's conclusions, genes A and B are 6 map units apart because they cross over 6 percent of the time. Similarly, genes B and C are 12.5 map units apart because they cross over 12.5 percent of the time. But where are the genes located in relation to each other on the chromosome?

Connecting CONCEPTS

Crossing Over Recall from **Chapter 6** that segments of non-sister chromatids can be exchanged during meiosis.

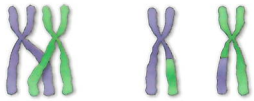
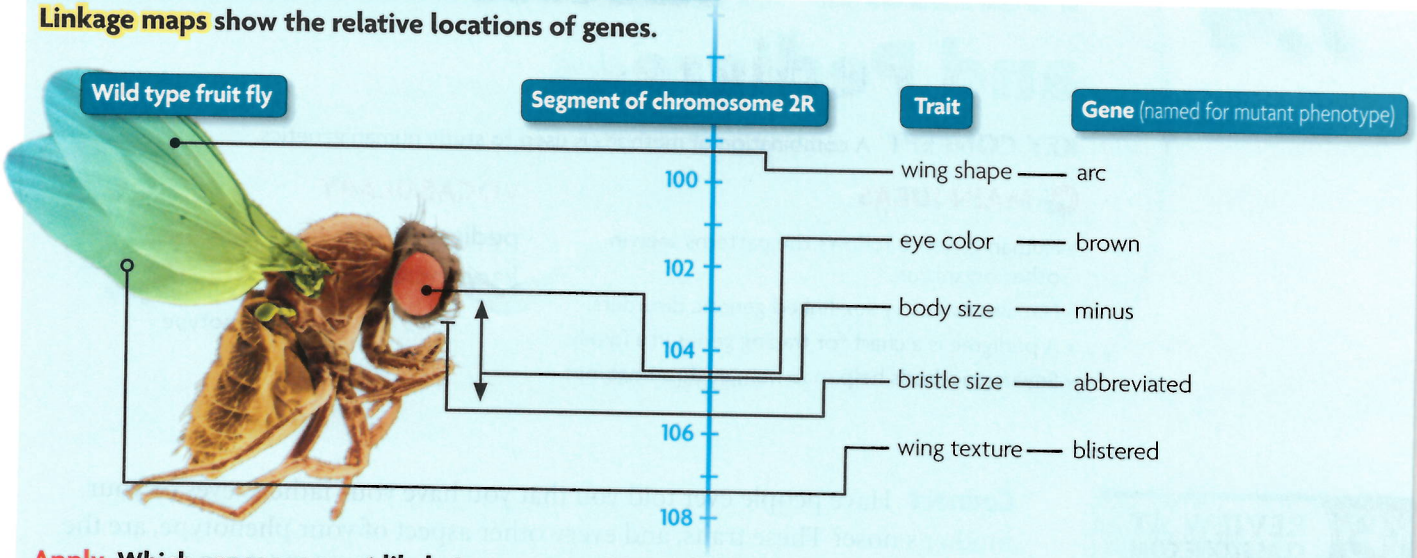


FIGURE 7.10 Gene Linkage in *Drosophila*

Linkage maps show the relative locations of genes.



Apply **Which genes are most likely to cross over? Least likely? Why?**

(colored SEM; magnification 20×)

Think about gene A as a point on a line. Gene B is either to the left or to the right of gene A. The same is true of the relationship between genes B and C. But if you only know the distances between genes A and B, and between genes B and C, you cannot determine the order of all three genes. You must also know the distance between genes A and C. As shown in **FIGURE 7.11**, the map distances between genes A and B and between genes B and C equal the map distance between genes A and C. Therefore, gene B must be located between genes A and C. If the map distance between genes A and C were 6.5 map units instead of 18.5 map units, then gene A would be between genes B and C.

Although linkage maps show the relative locations of linked genes, the maps do not show actual physical distances between genes. Linkage maps can give you a general idea about distances between genes, but many factors affect gene linkage. As a result, two pairs of genes may be the same number of map units apart, but they may not have the same physical distance between them.

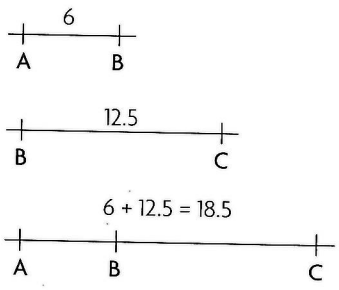


FIGURE 7.11 The order of genes on a chromosome can be determined if all of their cross-over frequencies are known.

Summarize **How can a linkage map be made from observations of traits?**

7.3 ASSESSMENT

ONLINE QUIZ ClassZone.com

REVIEWING MAIN IDEAS

1. Summarize the importance of comparing wild type and mutant fruit flies in genetic research.
2. How is a **linkage map** related to cross-overs that take place during meiosis?

CRITICAL THINKING

3. **Compare and Contrast** How are linked genes similar to sex-linked genes? How are they different?
4. **Apply** Draw a linkage map based on the following cross-over percentages for three gene pairs: A – B = 8%, B – C = 10%, and A – C = 2%.

Connecting CONCEPTS

5. **Scientific Process** Punnett, Bateson, and Morgan found phenotype ratios that differed from Mendel's results. Explain how these differences led to new hypotheses and new investigations in genetics.

7.4 Human Genetics and Pedigrees

KEY CONCEPT A combination of methods is used to study human genetics.

MAIN IDEAS

- Human genetics follows the patterns seen in other organisms.
- Females can carry sex-linked genetic disorders.
- A pedigree is a chart for tracing genes in a family.
- Several methods help map human chromosomes.

VOCABULARY

pedigree, p. 214
karyotype, p. 217

Review
phenotype, allele, sex-linked gene, genotype

REVIEW AT CLASSZONE.COM

Connect Have people ever told you that you have your father's eyes or your mother's nose? These traits, and every other aspect of your phenotype, are the result of the genes that you inherited from your parents. Which parts of your phenotype come from which parent? In some cases, such as hair color or eye color, it may be very easy to tell. Often, however, it is not so obvious.

MAIN IDEA

Human genetics follows the patterns seen in other organisms.

Fruit flies and pea plants may seem boring and simple, but the basic principles of genetics were worked out using those organisms. Humans follow the same patterns of heredity. First, meiosis independently assorts chromosomes when gametes are made for sexual reproduction. Second, human heredity involves the same relationships between alleles—dominant and recessive interactions, polygenic traits, and sex-linked genes, among others.

The inheritance of many traits is very complex. A single trait may be controlled by several genes that interact. As you read in Section 7.2, eye color is controlled by at least three different genes. And, although several genes affect height, a person's environment during growth and development plays a large role in his or her adult height. What might seem like an obvious phenotype is rarely as simple as it looks.

FIGURE 7.12 The widow's peak, or pointed hairline, is a phenotype produced by a dominant autosomal gene.

Nonetheless, single-gene traits are very helpful in understanding human genetics. One such trait is the shape of a person's hairline. A hairline with a downward point, such as a widow's peak shown in **FIGURE 7.12**, is a dominant trait. A straight hairline is a recessive trait. The inheritance of this trait follows the same dominant and recessive pattern as the traits in Mendel's pea plants. Many genetic disorders, such as Huntington's disease, hemophilia, and Duchenne's muscular dystrophy, are also caused by single genes that follow a dominant and recessive pattern. In fact, much of what is known about human genetics comes from studying genetic disorders.

Apply **Why can the genetics of pea plants and fruit flies be applied to humans?**

MAIN IDEA

Females can carry sex-linked genetic disorders.

Recall from Section 7.1 that some genetic disorders are caused by autosomal genes. A carrier of an autosomal disorder does not show the disease but can pass on the disease-causing allele. Both males and females can be carriers of an autosomal disorder.

In contrast, only females can be carriers of sex-linked disorders. Several genetic disorders are caused by genes on the X chromosome, as you can see in **FIGURE 7.13.** Recall that males have an XY genotype. A male who has a gene for a disorder located on the X chromosome will not have a second, normal allele to mask it. One copy of the allele is enough for males to have the disorder. There are no male carriers of sex-linked disorders, because any male who has the gene displays the phenotype. Females can be carriers, because they may have a normal allele that gives them a normal phenotype. The likelihood of inheriting a sex-linked disorder depends both on the sex of the child and on which parent carries the disorder-causing allele. If only the mother has the allele and is a carrier, a child has a 50 percent chance of inheriting the allele. A daughter who inherits it will not show the phenotype, but a son will.

The British royal family provides a historical example of a sex-linked disorder. Queen Victoria (1819–1901) was a carrier of a recessive sex-linked allele for a disorder called hemophilia, which is a lack of proteins needed for blood to clot. People with hemophilia do not stop bleeding easily. Queen Victoria passed the allele to her son, who had hemophilia. He passed it to his daughter, who was a carrier, and so on. Members of royal families tended to marry into royal families in other countries, and by the early 1900s the royal families of several countries, including Russia and Spain, also had the allele for hemophilia. The allele in all of these people is traced back to Queen Victoria.

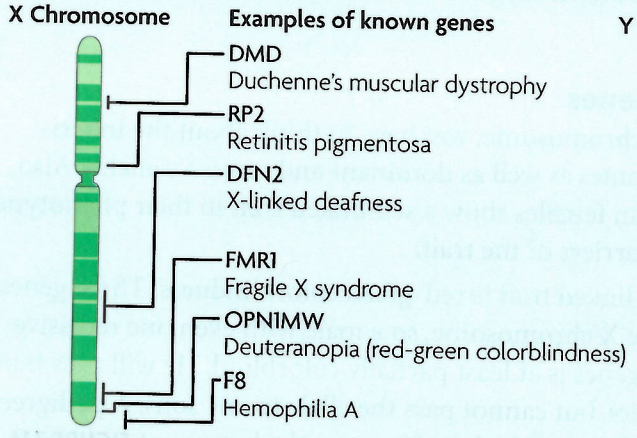
Contrast **How can carriers differ between autosomal and sex-linked disorders?**

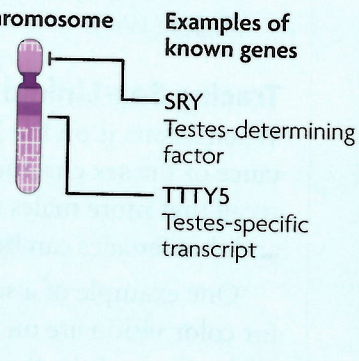
VOCABULARY

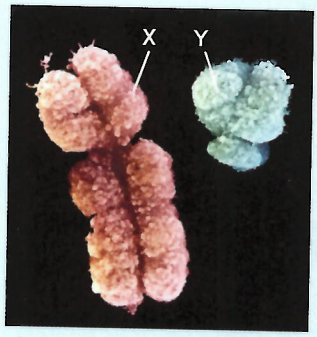
The term *carrier* means "a person who transports something." In genetics, a carrier is a person who "transports" a recessive allele but does not express the recessive phenotype.

FIGURE 7.13 Comparing the X and Y Chromosomes

The X chromosome has about 1100 known genes, including many that cause genetic disorders. The Y chromosome is about one-third the size of the X and has only about 250 known genes.







These X and Y chromosomes are duplicated and condensed. (colored SEM; magnification about 15,000×)

MAIN IDEA

A pedigree is a chart for tracing genes in a family.

Connecting CONCEPTS

Genetic Screening DNA testing is a direct method of studying genetic disorders. You will learn more about DNA tests and genetic screening in **Chapter 9.**

If two people want to know their child's chances of having a certain genetic disorder, they cannot rely upon their phenotypes. The parents also need to know their genotypes. A **pedigree** chart can help trace the phenotypes and genotypes in a family to determine whether people carry recessive alleles. When enough family phenotypes are known, genotypes can often be inferred.

A human pedigree shows several types of information. Boxes represent males and circles represent females. A shaded shape means that a person shows the trait, a white shape means that the person does not, and a shape that is half-shaded and half-white means that a person is a carrier. Lines connect a person to his or her mate, and to their children.

Using phenotypes to figure out the possible genotypes in a family is like putting pieces of a puzzle together. You have to use clues and logic to narrow the possibilities for each person's genotype. One particular clue, for example, can tell you whether the gene is on an autosome or on a sex chromosome. If approximately the same number of males and females have the phenotype, then the gene is most likely on an autosome. If, however, the phenotype is much more common in males, then the gene is likely on the X chromosome.

Tracing Autosomal Genes

It is fairly easy to trace genotypes through a pedigree when you know that you are dealing with a trait controlled by an autosomal gene. Why? A person who does not show the phenotype must have a homozygous recessive genotype. Any other genotype—either heterozygous or homozygous dominant—would produce the phenotype. Use the following steps to work your way through a pedigree for a gene on an autosome. The inheritance of an autosomal trait, such as the widow's peak described earlier, is shown on the top of **FIGURE 7.14.**

- People with a widow's peak have either homozygous dominant (*WW*) or heterozygous (*Ww*) genotypes.
- Two parents without a widow's peak are both homozygous recessive (*ww*), and cannot have children who have a widow's peak.
- Two parents who both have a widow's peak can have a child who does not (*ww*) if both parents are heterozygous for the dominant and recessive alleles (*Ww*).

Tracing Sex-Linked Genes

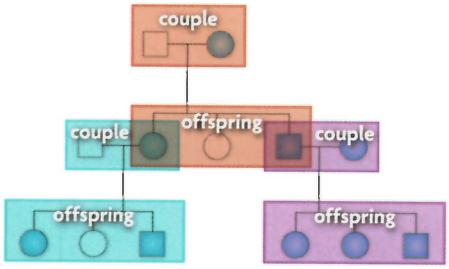
When a gene is on the X chromosome, you have to think about the inheritance of the sex chromosomes as well as dominant and recessive alleles. Also, recall that more males than females show a sex-linked trait in their phenotype, and that females can be carriers of the trait.

One example of a sex-linked trait is red-green colorblindness. Three genes for color vision are on the X chromosome, so a male with even one recessive allele of one of the three genes is at least partially colorblind. He will pass that allele to all of his daughters, but cannot pass the allele to any sons. A pedigree for colorblindness, which is sex-linked, is shown on the bottom of **FIGURE 7.14.**

FIGURE 7.14 Interpreting Pedigree Charts

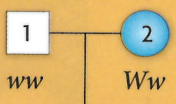
Figuring out genotypes from phenotypes requires you to use a process of elimination. You can often determine which genotypes are possible, and which ones are not.

Male without phenotype
Male with phenotype
Male carrier
Female without phenotype
Female with phenotype
Female carrier



TRACING AUTOSOMAL GENES: WIDOW'S PEAK

Parental generation



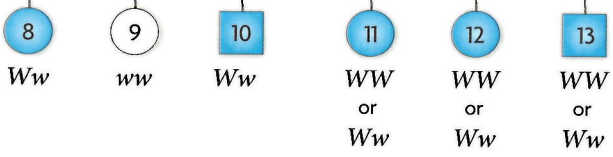
W = Dominant
w = Recessive

- Male 1 must be ww and female 2 must be heterozygous (Ww), because they have a daughter (5) with the recessive trait.

F_1 generation

- Children 4 and 6 have the widow's peak trait. They must be heterozygous, because they can inherit only one dominant allele.

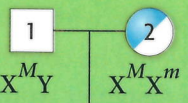
F_2 generation



- Children 8 and 10 have the widow's peak trait. They must be heterozygous, because they can inherit only one dominant allele.

TRACING SEX-LINKED GENES: COLORBLINDNESS

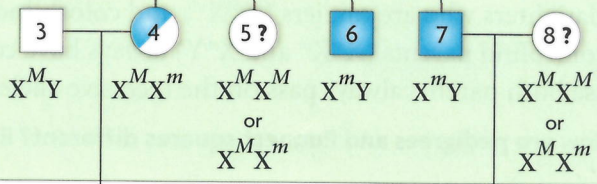
Parental generation



X^M = Dominant
X^m = Recessive

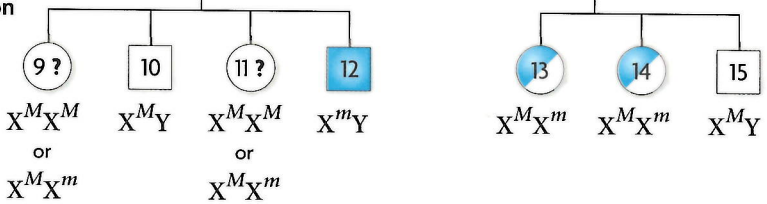
- Male 1 must be X^MY and female 2 must be a carrier (X^MX^m) because they have two colorblind sons.

F_1 generation



- Female 4 must be a carrier (X^MX^m) because she has a colorblind son. Males 6 and 7 must be X^mY. Females 5 and 8 are not colorblind, but it is not possible to determine whether they are carriers.

F_2 generation



- Children 13 and 14 must be carriers because their father is colorblind. Females 9 and 11 are not colorblind, but it is not possible to determine whether they are carriers.

CRITICAL VIEWING **Explain why it is not possible to identify all of the genotypes in the pedigree charts above. What information would you need to identify the genotypes of those people?**

FIGURE 7.15 RED-GREEN COLORBLINDNESS

A person with normal color vision can easily distinguish between different colors. A person who is red-green colorblind cannot.

PHENOTYPE	GENOTYPES	PHENOTYPE	GENOTYPES
normal vision	X^MX^m *or* X^MX^M *or* X^MY	red-green colorblind	X^mX^m *or* X^mY

The steps below can be applied to any sex-linked trait. By using a process of elimination, you can often figure out the possible genotypes for a given phenotype. First, think about the individuals shown in the pedigree chart.

- Colorblind females must be homozygous recessive (X^mX^m).
- Males who are colorblind must have the recessive allele (X^mY).
- Females who are heterozygous for the alleles (X^MX^m) do not show the phenotype, but they are carriers of the trait.

Then think about the possible offspring of the people shown in the pedigree.

- A female carrier (X^MX^m) and a male with normal color vision (X^MY) have a 50 percent chance that a son would be colorblind (X^mY). The same couple has a 50 percent chance that a daughter would be a carrier (X^MX^m).
- Colorblind females (X^mX^m) and males with normal color vision (X^MY) will have daughters who are carriers (X^MX^m) and colorblind sons (X^mY).
- Two colorblind parents (X^mX^m and X^mY) always have colorblind children because both parents always pass on the recessive allele.

Contrast **How are pedigrees and Punnett squares different? Explain.**

MAIN IDEA

Several methods help map human chromosomes.

The human genome, or all of the DNA in a human cell, is so large that mapping human genes is difficult. As a result, a combination of several methods is used. Pedigrees are useful for studying genetics in a family. Scientists can even gather a large number of pedigrees from people who are not related to look for inheritance patterns.

Other methods more directly study human chromosomes. A **karyotype** (KAR-ee-uh-TYP), for example, is a picture of all of the chromosomes in a cell. In order to study the chromosomes, chemicals are used to stain them. The chemical stains produce a pattern of bands on the chromosomes, as shown in **FIGURE 7.16.** The sizes and locations of the bands are very consistent for each chromosome, but the bands differ greatly among different chromosomes. Therefore, different chromosomes can be easily identified in a karyotype.

Karyotypes can show changes in chromosomes. Chromosome changes can be dramatic, such as when a person has too many chromosomes. In Down syndrome, for example, a person has an extra copy of at least part of chromosome 21. In XYY syndrome, a male has an extra Y chromosome. Other times, a karyotype reveals the loss of part of a chromosome. In the figure, you can see a deletion of a large part of chromosome 1. Scientists also use karyotypes to estimate the distances between genes on a chromosome. A karyotype can help show the possible location of a gene on a chromosome.

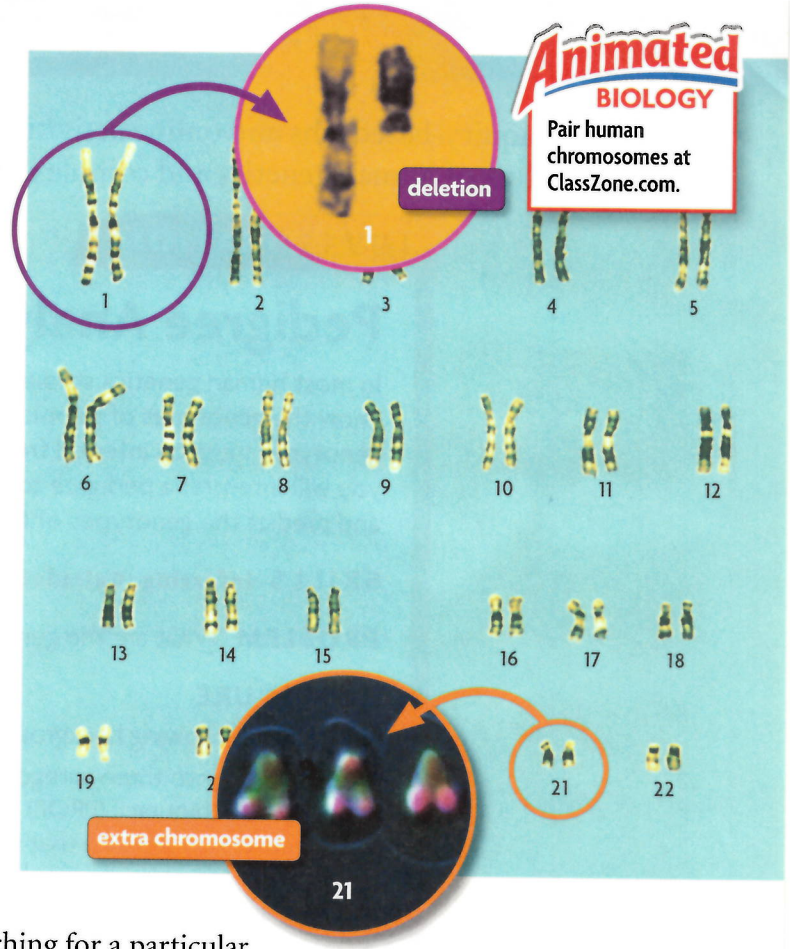


FIGURE 7.16 A karyotype can help show chromosomal disorders, such as the deletion in chromosome 1 (top inset) and the extra chromosome 21 in Down syndrome (bottom inset). (LM; magnifications: deletion 8000×; colored LM, extra chromosome 11,000×)

Chromosome mapping can be done directly by searching for a particular gene. All of the chromosomes are cut apart into smaller pieces. Then this library of chromosome parts is searched to find the gene. Although many genes and their locations have been identified through this process, it is a slow and inefficient method. The large-scale mapping of all of the genes on human chromosomes truly began with the Human Genome Project, which you will read more about in Chapter 9.

Apply **Why must a combination of methods be used to study human genetics?**

7.4 ASSESSMENT

ONLINE QUIZ ClassZone.com

REVIEWING MAIN IDEAS

1. How can Mendel's principles be used to study human traits?
2. Is a person who is homozygous recessive for a recessive genetic disease a carrier? Explain.
3. Describe how phenotypes can be used to predict genotypes in a **pedigree.**
4. What is a **karyotype,** and how can it be used to study human chromosomes?

CRITICAL THINKING

5. **Apply** Suppose a colorblind male and a female with no recessive alleles for colorblindness have children. What is the probability they will have a colorblind son? a colorblind daughter?
6. **Contrast** How do pedigrees for autosomal genes differ from pedigrees for sex-linked genes?

Connecting CONCEPTS

7. **Principles of Genetics** Explain why Mendel's principles of inheritance can be applied to all sexually reproducing species.

Use these inquiry-based labs and online activities to deepen your understanding of human genetics and complex patterns of inheritance.

INVESTIGATION

Pedigree Analysis

In most human genetics studies, scientists do not know the genotypes of people involved, so possible genotypes must be inferred from pedigrees. In this lab, you will interpret a pedigree to determine genotypes and predict the genotypes of future offspring.

MATERIALS
Pedigree Datasheet

SKILLS Inferring, Calculating Probabilities

PROBLEM What are the genotypes of the people in the pedigree?

PROCEDURE

1. Read the following background information and pedigree.

   People fall into three categories for the ability to taste a bitter chemical called 6-n-propylthiouracil (PROP). People who can taste PROP find it very unpleasant. Scientists hypothesize that these people, called supertasters, are homozygous for the trait (T_1 T_1). People who are heterozygous (T_1 T_2), called medium tasters, taste PROP as being somewhat bitter. Nontasters (T_2 T_2) do not taste the bitterness at all.

2. On your datasheet, fill in the possible genotypes for each person, including the phenotypes for those people who are medium tasters.

ANALYZE AND CONCLUDE

1. **Calculate** What is the probability that Jack will be a supertaster? What is the probability that Jill will be a supertaster? Explain your answers.
2. **Analyze** Is the gene for being a supertaster autosomal or sex-linked? Explain your answer based on the pedigree chart.