Patterns of Heredity and Human Genetics

What You’ll Learn

- You will compare the inheritance of recessive and dominant traits in humans.
- You will analyze the inheritance of incompletely dominant and codominant traits.
- You will determine the inheritance of sex-linked traits.

Why It’s Important

The transmission of traits from generation to generation affects your appearance, your behavior, and your health. Understanding how these traits are inherited is important in understanding the traits you may pass on to a future generation.

Before starting the chapter, make a list of some physical characteristics that may appear in your family members. As you read the chapter, note whether your family members have dominant or recessive traits discussed in the text.

To find out more about human genetics, visit the Glencoe Science Web site. science.glencoe.com

It is difficult to imagine how the information for such varied traits as eye or hair color and athletic talent could be contained in the nucleic acids composing this chromosome.
Making a Pedigree

At some point, you have probably seen a family tree, either for your family or for someone else’s. A family tree traces a family name and various family members through successive generations. Through a family tree, you can trace your cousins, aunts, uncles, grandparents, and great-grandparents.

Pedigrees illustrate inheritance

Geneticists often need to map the inheritance of genetic traits from generation to generation. A pedigree is a graphic representation of genetic inheritance. At a glance, it looks very similar to any family tree.

A pedigree is made up of a set of symbols that identify males and females, individuals affected by the trait being studied, and family relationships. Some commonly used symbols are shown in Figure 12.1. A circle represents a female; a square represents a male.

Figure 12.1
Geneticists use these symbols to make and analyze a pedigree.
represents a male. Shaded circles and squares represent individuals showing the trait being studied. Unshaded circles and squares designate individuals that do not show the trait. A half-shaded circle or square represents a carrier, a heterozygous individual. A horizontal line connecting a circle and a square indicates that the individuals are parents, and a vertical line connects a set of parents with their offspring. Each horizontal row of circles and squares in a pedigree designates a generation, with the most recent generation shown at the bottom. The generations are identified in sequence by Roman numerals, and each individual is given an Arabic number. You can practice using these symbols to make a pedigree in the MiniLab on this page.

Analyzing a pedigree

An example of a pedigree for a fictitious rare, recessive disorder in humans is shown in Figure 12.2. This disorder could be any of several recessive disorders in which the disorder shows up only if the affected person carries two recessive alleles for the trait. Follow the pedigree as you read how to analyze it.

Suppose individual III-1 in the pedigree wants to know the likelihood of passing on this allele to her children. By studying the pedigree, the individual will be able to determine the likelihood that she carries the allele. Notice that information can also be gained about other members of the family by studying the pedigree. For example, you know that I-1 and I-2 are both carriers of the recessive allele for the trait because they have produced II-3, who shows the recessive phenotype. If you drew a Punnett square for the mating of individuals I-1 and I-2, you...
would find, according to Mendelian segregation, that the ratio of homozygous dominant to heterozygous to homozygous recessive genotypes among their children would be 1:2:1. Of those genotypes possible for the members of generation II, only the homozygous recessive genotype will express the trait, which is the case for II-3.

You can’t tell the genotypes of II-4 and II-5, but they have a normal phenotype. If you look at the Punnett square you made, you can see that the probability of II-4 and of II-5 being a carrier is each two out of three because they can have only two possible genotypes—homozygous normal and heterozygous. The homozygous recessive genotype is not a possibility in these individuals because neither of them shows the affected phenotype.

Because none of the children in generation III are affected and because the recessive allele is rare, it is reasonably safe to assume that II-1 is not a carrier. You know that individual II-2 must be a carrier like her parents because she has passed on the recessive allele to subsequent generation IV. Because individual III-1 has one parent who is heterozygous and the other parent who is assumed to be homozygous normal, III-1 most likely has a one-in-two chance of being a carrier. If her parent II-1 had been heterozygous instead of homozygous normal, III-1’s chances of being a carrier are increased to two in three.

**Simple Recessive Heredity**

Most genetic disorders are caused by recessive alleles. Many of these alleles are relatively rare, but a few are common in certain ethnic groups. You can practice calculating the chance that offspring will be born with some of these genetic traits in the *Problem-Solving Lab* above.
Cystic fibrosis

Cystic fibrosis (CF) is a fairly common genetic disorder among white Americans. Approximately one in 20 white Americans carries the recessive allele, and one in 2000 children born to white Americans inherits the disorder. Due to a defective protein in the plasma membrane, cystic fibrosis results in the formation and accumulation of thick mucus in the lungs and digestive tract. Physical therapy, special diets, and new drug therapies have continued to raise the average life expectancy of CF patients.

Tay-Sachs disease

Tay-Sachs (tay saks) disease is a recessive disorder of the central nervous system. In this disorder, a recessive allele results in the absence of an enzyme that normally breaks down a lipid produced and stored in tissues of the central nervous system. Therefore, this lipid fails to break down properly and accumulates in the cells. The allele for Tay-Sachs is especially common in the United States among Ashkenazic Jews, whose ancestors came from eastern Europe. Figure 12.3 shows a typical pedigree for Tay-Sachs disease.

Phenylketonuria

Phenylketonuria (fen ul keet un YOOR ee uh), also called PKU, is a recessive disorder that results from the absence of an enzyme that converts one amino acid, phenylalanine, to a different amino acid, tyrosine. Because phenylalanine cannot be broken down, it and its by-products accumulate in the body and result in severe damage to the central nervous system. The PKU allele is most common among people whose ancestors came from Norway or Sweden.

A homozygous PKU newborn appears healthy at first because its mother’s normal enzyme level prevented phenylalanine accumulation during development. However, once the infant begins drinking milk, which is rich in phenylalanine, the amino acid accumulates and mental retardation occurs. Today, a PKU test is normally performed on all infants a few days after birth. Infants affected by PKU are given a diet that is low in phenylalanine until their brains are fully developed. With this special diet, the toxic effects of the disorder can be avoided.

Ironically, the success of treating phenylketonuria infants has resulted in a new problem. If a female who is homozygous recessive for PKU becomes pregnant, the high phenylalanine levels in her blood can damage her fetus—the developing baby. This problem occurs even if the fetus is heterozygous and would be phenotypically normal. You may have
noticed PKU warnings on cans of diet soft drinks. Because most diet drinks are sweetened with an artificial sweetener that contains phenylalanine, a pregnant woman who is homozygous recessive must limit her intake of diet foods.

**Simple Dominant Heredity**

Unlike the inheritance of recessive traits in which a recessive allele must be inherited from both parents for a person to show the recessive phenotype, many traits are inherited just as the rule of dominance predicts. Remember that in Mendelian inheritance, a single dominant allele inherited from one parent is all that is needed for a person to show the dominant trait.

**Simple dominant traits**

Tongue rolling is one example of a simple dominant trait. If you can roll your tongue, you’ve inherited the dominant allele from at least one of your parents. A Hapsburg lip is shown in Figure 12.4 along with earlobe types, another dominant trait that is determined by simple Mendelian inheritance. Having earlobes that are attached to the head is a recessive trait (ff), whereas heterozygous (Ff) and homozygous dominant (FF) individuals have earlobes that hang freely.

There are many other human traits that are inherited by simple dominant inheritance. Figure 12.5 shows one of these traits—hitchhiker’s thumb, the ability to bend your thumb tip.
### Understanding Main Ideas

1. In your own words, define the following symbols used in a pedigree: a square, a circle, an unshaded circle, a shaded square, a horizontal line, and a vertical line.

2. Describe one genetic disorder that is inherited as a recessive trait.

3. How are the cause and onset of symptoms of Huntington’s disease different from those of PKU and Tay-Sachs disease?

4. Describe one trait that is inherited as a dominant allele. If you carried that trait, would you necessarily pass it on to your children?

### Huntington’s disease

Huntington’s disease is a lethal genetic disorder caused by a rare dominant allele. It results in a breakdown of certain areas of the brain. No effective treatment exists.

Ordinarily, a dominant allele with such severe effects would result in death before the affected individual could have children and pass the allele on to the next generation. But because the onset of Huntington’s disease usually occurs between the ages of 30 and 50, an individual may have children before knowing whether he or she carries the allele. A genetic test has been developed that allows individuals to check their DNA. Although this test allows carriers to decide whether they want to have children and risk passing the trait on to future generations, it also places a tremendous burden on them in knowing they will develop the disease. For this reason, some people may choose not to be tested. The pedigree in Figure 12.6 shows a typical pattern of occurrence of Huntington’s disease in a family.

Notice that every child of an affected individual has a 50 percent chance of being affected and then a 50 percent chance of passing the defective allele to his or her own child.

![Pedigree](image)

**Figure 12.6**

This is a typical pedigree for a simple, dominant inheritance of Huntington’s disease. This particular chart shows the disorder in each generation and equally distributed among males and females.

backward more than 30 degrees. A straight thumb is recessive. Other dominant traits in humans include almond-shaped eyes (round eyes are recessive), thick lips (thin lips are recessive), and the presence of hair on the middle section of your fingers.

### Section Assessment

#### Thinking Critically

5. Suppose that a child with free-hanging earlobes has a mother with attached earlobes. Can a man with attached earlobes be the child’s father?

6. Interpreting Scientific Illustrations  Make a pedigree for three generations of a family that shows at least one member of each generation who demonstrates a particular trait. Would this trait be dominant or recessive? For more help, refer to Thinking Critically in the Skill Handbook.
Variations in the pattern of inheritance explained by Mendel became known soon after his work was discovered. What do geneticists do when observed patterns of inheritance, such as kernel color in this ear of corn, do not appear to follow Mendel’s laws? They often use a strategy of piecing together bits of a puzzle until the basis for the unfamiliar inheritance pattern is understood.

Complex Patterns of Inheritance

Patterns of inheritance that are explained by Mendel’s experiments are often referred to as simple Mendelian inheritance—the inheritance controlled by dominant and recessive paired alleles. However, many inheritance patterns are more complex than those studied by Mendel. As you will learn, most traits are not simply dominant or recessive. The BioLab at the end of this chapter investigates a type of inheritance that doesn’t even involve chromosomes.

Incomplete dominance: Appearance of a third phenotype

When inheritance follows a pattern of dominance, heterozygous and homozygous dominant individuals both have the same phenotype. When traits are inherited in an incomplete dominance pattern, however, the phenotype of the heterozygote is intermediate between those of the two homozygotes. For example, if a homozygous red-flowered snapdragon plant (RR) is crossed with a homozygous white-flowered snapdragon plant (R’R’), all of the F₁ offspring will have pink

The genetics of Indian corn (above) is often like a puzzle (inset).
flowers, as shown in Figure 12.7. The intermediate pink form of the trait occurs because neither allele of the pair is completely dominant. Note that the letters \( R \) and \( R' \), rather than \( R \) and \( r \), are used to show that these alleles are incompletely dominant.

The new phenotype occurs because the flowers contain a colored pigment. The \( R \) allele codes for an enzyme that produces a red pigment. The \( R' \) allele codes for a defective enzyme that makes no pigment. Because the heterozygote has only one copy of the \( R \) allele, its flowers produce only half the amount of red pigment that the flowers of the red homozygote produce, and they appear pink. The \( R'R' \) homozygote has no normal enzyme, produces no red pigment, and appears white.

Note that the segregation of alleles is the same as in simple Mendelian inheritance. However, because neither allele is dominant, the plants of the \( F_1 \) generation all have pink flowers. When pink-flowered \( F_1 \) plants are crossed with each other, the offspring in the \( F_2 \) generation appear in a 1:2:1 phenotypic ratio of red to pink to white flowers. This result supports Mendel’s law of independent assortment, which states that the alleles are inherited independently.

**Codominance: Expression of both alleles**

In chickens, black-feathered and white-feathered birds are homozygotes for the \( B \) and \( W \) alleles, respectively. Two different uppercase letters are used to represent the alleles in codominant inheritance.

One of the resulting heterozygous offspring in a breeding experiment between a black rooster and a white hen is shown in Figure 12.8. You might expect that heterozygous chickens, \( BW \), would be black if the pattern of inheritance followed Mendel’s law of dominance, or gray if the trait were
incompletely dominant. Notice, however, that the heterozygote is neither black nor gray. Instead, all of the offspring are checkered; some feathers are black and other feathers are white. In such situations, the inheritance pattern is said to be codominant. **Codominant alleles** cause the phenotypes of both homozygotes to be produced in heterozygous individuals. In codominance, both alleles are expressed equally.

**Multiple phenotypes from multiple alleles**

Although each trait has only two alleles in the patterns of heredity you have studied thus far, it is common for more than two alleles to control a trait in a population. This is understandable when you recall that a new allele can be formed any time a mutation occurs in a nitrogen base somewhere within a gene. Although only two alleles of a gene can exist within a diploid cell, multiple alleles for a single gene can be studied in a population of organisms.

Traits controlled by more than two alleles have **multiple alleles**. The pigeons pictured in **Figure 12.9** show the effects of multiple alleles for feather color. Three alleles of a single gene govern their feather color, although each pigeon can have only two of these alleles. The number of alleles for any particular trait is not limited to three, and there are instances in which more than 100

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**Figure 12.9**

In pigeons, a single gene that controls feather color has three alleles. An enzyme that activates the production of a pigment is controlled by the **B** allele. This enzyme is lacking in **bb** pigeons.

- **A** The dominant **B** allele produces ash-red colored feathers.
- **B** The **B** allele produces wild-type blue feathers. **B** is dominant to **b** but recessive to **B**.
- **C** The allele **b** produces a chocolate-colored feather and is recessive to both other alleles.
alleles are known to exist for a single trait! You can learn about another example of multiple alleles in the Problem-Solving Lab shown here.

**Sex determination**

Recall that in humans the diploid number of chromosomes is 46, or 23 pairs. There are 22 pairs of matching homologous chromosomes called **autosomes**. Homologous autosomes look exactly alike. The 23rd pair of chromosomes differs in males and females. These two chromosomes, which determine the sex of an individual, are called **sex chromosomes**. In humans, the chromosomes that control the inheritance of sex characteristics are indicated by the letters X and Y. If you are a human female, XX, your 23rd pair of chromosomes are homologous and look alike, as shown in Figure 12.10A. However, if you are a male, XY, your 23rd pair of chromosomes look different. Males, which have one X and one Y chromosome, produce two kinds of gametes, X and Y, by meiosis. Females have two X chromosomes, so they produce only X gametes. Figure 12.10B shows that after fertilization, a 1:1 ratio of males to females is expected. Because fertilization is governed by the laws of

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**Table 12.2 Coat color in rabbits**

<table>
<thead>
<tr>
<th>Phenotype</th>
<th>Allele</th>
<th>Pattern of inheritance</th>
</tr>
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<tbody>
<tr>
<td>Dark gray coat</td>
<td>C</td>
<td>dominant to all other alleles</td>
</tr>
<tr>
<td>Chinchilla</td>
<td>c&lt;sup&gt;h&lt;/sup&gt;</td>
<td>dominant to Himalayan and to white</td>
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<tr>
<td>Himalayan</td>
<td>c</td>
<td>dominant to white</td>
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<tr>
<td>White</td>
<td>c</td>
<td>recessive</td>
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**Thinking Critically**

1. List all possible genotypes for a
   a. dark gray-coated rabbit (there are 4).
   b. chinchilla rabbit (there are 3).
   c. Himalayan rabbit (there are 2).
   d. white rabbit (there is 1).

2. Predict the phenotype for a rabbit with a c<sup>h</sup>c<sup>h</sup> and with a Cc<sup>h</sup> genotype. Explain your answer.

3. Would it be possible to obtain white rabbits if one parent is white and the other is chinchilla? Explain your answer.

4. Would it be possible to obtain chinchilla rabbits if one parent is Himalayan and the other is white? Explain.

5. A chinchilla rabbit is mated with a Himalayan. Some offspring are white. What are the parents’ genotypes?
probability, the ratio usually is not exactly 1:1 in a small population.

**Sex-linked inheritance**

*Drosophila* (droh SAHF uh luH), commonly known as fruit flies, inherit sex chromosomes in the same way as humans do. Traits controlled by genes located on sex chromosomes are called sex-linked traits. The alleles for sex-linked traits are written as superscripts of the X or Y chromosome. Because the X and Y chromosomes are not homologous, the Y chromosome has no corresponding allele to one on the X chromosome and no superscript is used. Also remember that any allele on the X chromosome of a male will not be masked by a corresponding allele on the Y chromosome.

In 1910, Thomas Hunt Morgan discovered traits linked to sex chromosomes. Morgan noticed one day that one male fly had white eyes rather than the usual red eyes. He crossed the white-eyed male with a homozygous red-eyed female. All of the F₁ offspring had red eyes, indicating that the white-eyed trait is recessive. Then Morgan allowed the F₁ flies to mate among themselves. According to simple Mendelian inheritance, if the trait were recessive, the offspring in the F₂ generation would show a 3:1 ratio of red-eyed to white-eyed flies. As you can see in Figure 12.11, this is what Morgan observed. However, he also noticed that the trait of white eyes appeared only in male flies.

Morgan hypothesized that the red-eye allele was dominant and the white-eye allele was recessive. He also reasoned that the gene for eye color was located on the X chromosome and was not present on the Y chromosome. In heterozygous females, the dominant allele for red eyes masks the recessive allele for white eyes. In males, however, a single recessive allele is expressed as a white-eyed phenotype. When Morgan crossed a heterozygous red-eyed female (X⁺X⁻) with a white-eyed male (X⁻Y), the F₁ offspring all had red eyes (X⁺X⁻ Y⁻ or X⁻ X⁺ Y⁻). The F₂ generation showed a 3:1 ratio of red-eyed to white-eyed flies, with all females having red eyes and half of the males having white eyes.

**Figure 12.11**

Morgan crossed a white-eyed male fruit fly with a normal homozygous red-eyed female (a). He then allowed the F₁ flies to mate (b). The superscripts R and r are the dominant and recessive alleles for eye color in fruit flies.

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<tbody>
<tr>
<td>a</td>
<td>X⁺ Y⁻</td>
</tr>
<tr>
<td>b</td>
<td>X⁺ X⁺ Y⁻</td>
</tr>
</tbody>
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F₁ All red eyed

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<tr>
<td>F₂</td>
<td>Females: all red eyed</td>
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<td></td>
<td>Males: 1/2 red eyed 1/2 white eyed</td>
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Slide 12.11

Morgan crossed a white-eyed male fruit fly with a normal homozygous red-eyed female (a). He then allowed the F₁ flies to mate (b). The superscripts R and r are the dominant and recessive alleles for eye color in fruit flies.
female with a white-eyed male, half of all the males and half of all the females inherited white eyes. The only explanation of these results is Morgan’s hypothesis: The allele for eye color is carried on the X chromosome and the Y chromosome has no allele for eye color.

Traits dependent on genes that follow the inheritance pattern of a sex chromosome are called sex-linked traits. Eye color in fruit flies is an example of an X-linked trait. Y-linked traits are passed only from male to male.

### Polygenic inheritance

Some traits, such as skin color and height in humans, and cob length in corn, vary over a wide range. Such ranges occur because these traits are governed by many different genes. **Polygenic inheritance** is the inheritance pattern of a trait that is controlled by two or more genes. The genes may be on the same chromosome or on different chromosomes, and each gene may have two or more alleles. For simplicity, uppercase and lowercase letters are used to represent the alleles, as they are in Mendelian inheritance. Keep in mind, however, that the allele represented by an uppercase letter is not dominant. All heterozygotes are intermediate in phenotype.

In polygenic inheritance, each allele represented by an uppercase letter contributes a small, but equal, portion to the trait being expressed. The result is that the phenotypes usually show a continuous range of variability from the minimum value of the trait to the maximum value.

Suppose, for example, that stem length in a plant is controlled by three different genes: A, B, and C. Each gene is on a different chromosome and has two alleles, which can be represented by uppercase or lowercase letters. Thus, each diploid plant has a total of six alleles for stem length. A plant that is homozygous for short alleles at all three gene locations (aabbcc) might grow to be only 4 cm tall, the base height. A plant that is homozygous for tall alleles at all three gene locations (AABBCC) might be 16 cm tall. The difference between the tallest possible plant and the shortest possible plant is 12 cm, or 2 cm per each of the six tall alleles. You could say that each allele represented by an uppercase letter contributes 2 cm to the total height of the plant.

Suppose a 16-cm-tall plant were crossed with a 4-cm-tall plant. In the F₁ generation, all the offspring would be AaBbCc. If each of the three tall genes A, B, and C contributed 2 cm of height to the base height of 4 cm, the plants would be 10 cm tall (4 cm + 6 cm)—intermediate in height. If they are allowed to interbreed, the F₂ offspring

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**Figure 12.12**

In this example of polygenic inheritance, three genes each have two alleles that contribute to the trait. When the distribution of plant heights is graphed, a bell-shaped curve is formed. Intermediate heights occur most often.

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**Word Origin**

polygenic

From the Greek words polys, meaning “many,” and genos, meaning “kind.” Polygenic inheritance involves many genes.
will show a broad range of heights. A Punnett square of this trihybrid cross would show that 10-cm-tall plants are most often expected, and the tallest and shortest plants are seldom expected. Notice in Figure 12.12 that when these results are graphed, the shape of the graph confirms the prediction of the Punnett square.

Environmental Influences

Even when you understand dominance and recessiveness and you have solved the puzzles of the other patterns of heredity, the inheritance picture is not complete. The genetic makeup of an organism at fertilization determines only the organism’s potential to develop and function. As the organism develops, many factors can influence how the gene is expressed, or even whether the gene is expressed at all. Two such influences are the organism’s external and internal environments.

Influence of external environment

Sometimes, individuals known to have a particular gene fail to express the phenotype specified by that gene. Temperature, nutrition, light, chemicals, and infectious agents all can influence gene expression. In certain bacteria, temperature has an effect on the expression of color, as shown in Figure 12.13. External influences can also be seen in leaves. Leaves on a tree can have different sizes and shapes depending on the amount of light they receive.

Influence of internal environment

The internal environments of males and females are different because of hormones and structural differences, Figure 12.14. For example, traits such as horn size in mountain sheep, male-pattern baldness in humans, and feather color in peacocks are expressed differently in the genotypes are identical.
You can now see that you must learn how genes interact with each other and with the environment to form a more complete picture of inheritance. Mendel’s idea that heredity is a composite of many individual traits still holds. Later researchers have filled in more details of Mendel’s great contributions.

**Figure 12.15**

Some traits are expressed differently in the sexes.

A. The plumage of the male peacock is highly decorated and colored.

B. The plumage of the female peahen is dull by comparison.

C. Human male-pattern baldness, premature balding that occurs in a characteristic pattern, affects males but not females.

sexes, as you can see in Figure 12.15. These differences are controlled by different hormones, which are determined by different sets of genes.

An organism’s age also affects gene function. The nature of such a pattern is not well understood, but it is known that the internal environment of an organism changes with age.

### Understanding Main Ideas

1. A cross between a purebred animal with red hairs and a purebred animal with white hairs produces an animal that has both red hairs and white hairs. What type of inheritance pattern is involved?

2. In a cross between individuals of a species of tropical fish, all of the male offspring have long tail fins, and none of the females possess the trait. Mating of the F₁ fish fails to produce females with the trait. Explain a possible inheritance pattern of the trait.

3. A red-flowered sweet pea plant is crossed with a white-flowered sweet pea plant. All of the offspring are pink. What is the inheritance pattern being expressed?

4. The color of wheat grains shows a wide variability between red and white with multiple phenotypes. What type of inheritance pattern is being expressed?

### Thinking Critically

5. Armadillos always have four offspring that have identical genetic makeup. Suppose that, within a litter, each young armadillo is found to have a different phenotype for a particular trait. How could you explain this phenomenon?

6. **Forming a Hypothesis** An ecologist observes that a population of plants in a meadow has flowers that may be red, yellow, white, pink, or purple. Hypothesize what the inheritance pattern might be. For more help, refer to *Practicing Scientific Methods* in the Skill Handbook.
Codominance in Humans

Remember that in codominance, the phenotypes of both homozygotes are produced in the heterozygote. One example of this type of inheritance in humans is the disorder sickle-cell anemia.

Sickle-cell anemia

Sickle-cell anemia is a major health problem in the United States and in Africa. In the United States, it is most common in black Americans whose families originated in Africa and in white Americans whose families originated in the countries surrounding the Mediterranean Sea. About one in 12 African Americans, a much larger proportion than in most populations, is heterozygous for the disorder.

In an individual who is homozygous for the sickle-cell allele, the oxygen-carrying protein hemoglobin differs by one amino acid from normal hemoglobin. This defective hemoglobin forms crystal-like structures that change the shape of the red blood cells. The abnormal red blood cells are shaped like a sickle, or half-moon. The change in shape occurs in the body’s narrow capillaries after the hemoglobin releases oxygen to the cells. Abnormally shaped blood cells,
Figure 12.16, slow blood flow, block small vessels, and result in tissue damage and pain. Because sickled cells have a shorter life span than normal red blood cells, the person suffers from anemia, a condition in which there is a low number of red blood cells.

Individuals who are heterozygous for the allele produce both normal and sickled hemoglobin, an example of codominance. They produce enough normal hemoglobin that they do not have the serious health problems of those homozygous for the allele and can lead relatively normal lives. Individuals who are heterozygous are said to have the sickle-cell trait because they can show some signs of sickle-cell anemia if the availability of oxygen is reduced.

Multiple Alleles in Humans

Traits that are governed by simple Mendelian heredity have only two alleles. However, you have learned that more than two alleles of a gene are possible for certain traits. The ABO blood group is a classic example of a single gene that has multiple alleles in humans. How many alleles does this gene have? Read the Inside Story to answer this question.

Multiple alleles govern blood type

Human blood types, listed in Table 12.3, are determined by the presence or absence of certain molecules on the surfaces of red blood cells. As the determinant of blood types A, B, AB, and O, the gene I has three alleles: I^A, I^B, and i.

The importance of blood typing

Determining blood type is necessary before a person can receive a blood transfusion because the red blood cells of incompatible blood types could clump together, causing death. Blood typing can also be helpful in solving cases of disputed parentage. For example, if a child has type AB blood and its mother has type A, a man with type O blood could not possibly be the father. But blood tests cannot prove that a certain man definitely is the father; they indicate only that he could be. DNA tests are necessary to determine actual parenthood.
The ABO Blood Group

The gene for blood type, gene I, codes for a membrane protein found on the surface of red blood cells. Each of the three alleles codes for a different protein. Your immune system recognizes the red blood cells as belonging to you. If cells with a different protein enter your body, your immune system will attack them.

**Critical Thinking** If you inherit ii from your parents, what is your blood type?

1. **Phenotype A** The \( I^A \) allele is dominant to \( i \), so inheriting either the \( I^A i \) alleles or the \( I^A I^A \) alleles from your two parents will give you type A blood. Surface protein A is produced.

2. **Phenotype B** The \( I^B \) allele is also dominant to \( i \). To have type B blood, you must inherit the \( I^B \) allele from one parent and either another \( I^B \) allele or the \( i \) allele from the other. Surface protein B is produced.

3. **Phenotype AB** The \( I^A \) and \( I^B \) alleles are codominant to each other. This means that if you inherit the \( I^A \) allele from one parent and the \( I^B \) allele from the other, your red blood cells will produce both surface proteins and you will have type AB blood.

4. **Phenotype O** The \( i \) allele is recessive and produces no surface molecule. Therefore, if you are homozygous \( ii \), your blood cells have no surface proteins and you have blood type O.

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**Table 12.3 Human blood types**

<table>
<thead>
<tr>
<th>Genotypes</th>
<th>Surface Proteins</th>
<th>Phenotypes</th>
</tr>
</thead>
<tbody>
<tr>
<td>( I^A I^A ) or ( I^A i )</td>
<td>A</td>
<td>A</td>
</tr>
<tr>
<td>( I^B I^B ) or ( I^B i )</td>
<td>B</td>
<td>B</td>
</tr>
<tr>
<td>( I^A I^B )</td>
<td>A and B</td>
<td>AB</td>
</tr>
<tr>
<td>( ii )</td>
<td>none</td>
<td>O</td>
</tr>
</tbody>
</table>
Sex-Linked Traits in Humans

Several human traits are determined by genes that are carried on the sex chromosomes; most of these genes are located on the X chromosome. The pattern of sex-linked inheritance is explained by the fact that males, who are XY, pass an X chromosome to each of their daughters and a Y chromosome to each son. Females, who are XX, pass one of their X chromosomes to each child, Figure 12.17. If a son receives an X chromosome with a recessive allele from his mother, he will express the recessive phenotype because he has no chance of inheriting from his father a dominant allele that would mask the expression of the recessive allele.

Two traits that are governed by X-linked inheritance in humans are certain forms of color blindness and hemophilia. Determine whether Duchenne’s muscular dystrophy is sex-linked by reading the Problem-Solving Lab on this page.

Red-green color blindness

People who have red-green color blindness can’t differentiate these two...
colors. Red-green color blindness was first described in a boy who could not be trained to harvest only the ripe, red apples from his father’s orchard. Instead, he chose green apples as often as he chose red.

Other more serious problems can result from this disorder, such as the inability of color-blind people to identify red and green traffic lights by color. Color blindness is caused by the inheritance of either of two recessive alleles at two gene sites on the X chromosome that affect red and green receptors in the cells of the eyes.

**Hemophilia: An X-linked disorder**

Did you ever wonder about why a cut stops bleeding so quickly? This human adaptation is essential. If your blood didn’t have the ability to clot at all, any cut could take a long time to stop bleeding. Of greater concern would be internal bleeding resulting in a bruise, which a person may not immediately notice.

Hemophilia A is an X-linked disorder that causes just such a problem with blood clotting. About one male in every 10,000 has hemophilia, but only about one in 100 million females inherits the same disorder. Why? Males inherit the allele for hemophilia on the X chromosome from their carrier mothers. A single recessive allele for hemophilia will cause the disorder in males. Females would need two recessive alleles to inherit hemophilia. The family of Queen Victoria, pictured in the Social Studies Connection at the end of this chapter, is the best-known study of hemophilia A, also called royal hemophilia.

Hemophilia A can be treated with blood transfusions and injections of Factor VIII, the blood-clotting enzyme that is absent in people affected by the condition. However, both treatments are expensive. New methods of DNA technology are being used to develop a cheaper source of the clotting factor.

**Polygenic Inheritance in Humans**

Think of all the traits you inherited from your parents. Although many of your traits were inherited through simple Mendelian patterns or through multiple alleles, many other human traits are determined by polygenic inheritance. These kinds of traits usually represent a range of variation that is measurable. The MiniLab shown here examines one of these traits—the color variations in human eyes.
Skin color: A polygenic trait

In the early 1900s, the idea that polygenic inheritance occurs in humans was first tested using data collected on skin color. Scientists found that when light-skinned people mate with dark-skinned people, their offspring have intermediate skin colors. When these children produce the F2 generation, the resulting skin colors range from the light-skin color to the dark-skin color of the grandparents (the P1 generation), with most children having an intermediate skin color. As Figure 12.18 shows, the variation in skin color indicates that between three and four genes are involved.

Changes in Chromosome Numbers

You have been reading about traits that are caused by one or several genes on chromosomes. What would happen if an entire chromosome or part of a chromosome were missing from the complete set? What if cells had an extra chromosome? As you have learned, abnormal numbers of chromosomes usually, but not always, result from accidents of meiosis. Many abnormal phenotypic effects result from such mistakes.

Unusual numbers of autosomes

You know that a human usually has 23 pairs of chromosomes, or 46 chromosomes altogether. Of these 23 pairs of chromosomes, 22 pairs are autosomes. Humans who have an unusual number of autosomes all are trisomic—that is, they have three of a particular autosome instead of just two. In other words, they have 47 chromosomes. Recall that trisomy usually results from nondisjunction, which occurs when paired homologous chromosomes fail to separate properly during meiosis.

To identify an abnormal number of chromosomes, a sample of cells is obtained from an individual or from a fetus. Metaphase chromosomes are photographed, and the chromosome pictures are then enlarged, cut apart, and arranged in pairs on a chart.

Figure 12.18
This graph (b) shows the expected distribution of human skin color if controlled by one, three, or four genes. The observed distribution of skin color (a) closely matches the distribution shown by four genes.
according to length and location of the centromere, as Figure 12.19 shows. This chart of chromosome pairs is called a karyotype, and it is valuable in pinpointing unusual chromosome numbers in cells.

**Down syndrome: Trisomy 21**

Most disorders of chromosome number that occur in humans cause symptoms so severe that the developing fetus dies, often before the woman even realizes she is pregnant. Fortunately, these disorders occur only rarely. Down syndrome is the only autosomal trisomy in which affected individuals survive to adulthood. It occurs in about one in 700 live births.

Down syndrome is a group of symptoms that results from trisomy of chromosome 21. Individuals who have Down syndrome have at least some degree of mental retardation. The incidence of Down syndrome births is higher in older mothers, especially those over 40.

**Unusual numbers of sex chromosomes**

Many abnormalities in the number of sex chromosomes are known to exist. An X chromosome may be missing (designated as XO) or there may be an extra one (XXX or XXY). There may also be an extra Y chromosome (XYY), as you can see by examining Figure 12.19. Any individual with at least one Y chromosome is a male, and any individual without a Y chromosome is a female. Most of these individuals lead normal lives, but they cannot have children and some have varying degrees of mental retardation.

**Figure 12.19**

This karyotype demonstrates XYY syndrome, where two Y chromosomes are inherited in addition to an X chromosome.

### Section Assessment

#### Understanding Main Ideas

1. Why are sex-linked traits such as red-green color blindness and hemophilia more commonly found in males than in females? Explain your answer in terms of the X chromosome.
2. In addition to revealing chromosome abnormalities, what other information would a karyotype show?
3. What would the genotypes of parents have to be for them to have a color-blind daughter? Explain.
4. Describe a genetic trait in humans that is inherited as codominance. Describe the phenotypes of the two homozygotes and that of the heterozygote. Why is this trait an example of codominance?

#### Thinking Critically

5. A man is accused of fathering two children, one with type O blood and another with type A blood. The mother of the children has type B blood. The man has type AB blood. Could he be the father of both children? Explain your answer.

6. **Making and Using Tables**

   Construct a table of the traits discussed in this section. For column heads, use Trait, Pattern of inheritance, and Characteristics. For more help, refer to Organizing Information in the Skill Handbook.
What is the pattern of cytoplasmic inheritance?

The mitochondria of all eukaryotes and the chloroplasts of plants and algae contain DNA. This DNA is not coiled into chromosomes, but it still carries genes that control genetic traits. Many of the mitochondrial genes control steps in the respiration process.

The DNA in chloroplasts controls traits such as chlorophyll production. Lack of chlorophyll in some cells causes the appearance of white patches in a leaf. This trait is known as variegated leaf. In this BioLab, you will carry out an experiment to determine the pattern of cytoplasmic inheritance of the variegated leaf trait in *Brassica rapa*.

**Problem**
What inheritance pattern does the variegated leaf trait in *Brassica* show?

**Hypotheses**
Consider the possible evidence you could collect that would answer the problem question. Among the people in your group, form a hypothesis that you can test to answer the question, and write the hypothesis in your journal.

**Objectives**
In this BioLab, you will:
- **Determine** which crosses of *Brassica* plants will reveal the pattern of cytoplasmic inheritance.
- **Analyze** data from *Brassica* crosses.

**Possible Materials**
*Brassica rapa* seeds, normal and variegated potting soil and trays paintbrushes forceps single-edge razor blade light source labels

**Safety Precautions**
Always wear goggles in the lab. Handle the razor blade with extreme caution. Always cut away from you. Wash your hands with soap and water after working with plant material.

**Skill Handbook**
Use the *Skill Handbook* if you need additional help with this lab.
**Plan the Experiment**

1. Decide which crosses will be needed to test your hypothesis.
2. Keep the available materials in mind as you plan your procedure. How many seeds will you need?
3. Record your procedure, and list the materials and quantities you will need.
4. Assign a task to each member of the group. One person should write data in a journal, another can pollinate the flowers, while a third can set up the plant trays. Determine who will set up and clean up materials.
5. Design and construct a data table for recording your observations.

**Check the Plan**

Discuss the following points with other group members to decide the final procedure for your experiment.

1. What data will you collect, and how will data be recorded?
2. When will you pollinate the flowers? How many flowers will you pollinate?
3. How will you transfer pollen from one flower to another?
4. How and when will you collect the seeds that result from your crosses?
5. What variables will have to be controlled? What controls will be used?
6. When will you end the experiment?
7. **Make sure your teacher has approved your experimental plan before you proceed further.**
8. Carry out your experiment.

**Analyze and Conclude**

1. **Checking Your Hypothesis** Did your data support your hypothesis? Why or why not?
2. **Interpreting Observations** What is the inheritance pattern of variegated leaves in *Brassica*?
3. **Making Inferences** Explain why genes in the chloroplast are inherited in this pattern.
4. **Drawing Conclusions** Which parent is responsible for passing the variegated trait to its offspring?
5. **Making Scientific Illustrations** Draw a diagram tracing the inheritance of this trait through cell division.

**Going Further**

**Project** Make crosses between normal *Brassica* plants and genetically dwarfed, mutant *Brassica* plants to determine the inheritance pattern of the dwarf mutation.

**Biology Online** To find out more about inheritance of traits, visit the Glencoe Science Web site.

science.glencoe.com
One of the most famous examples of a pedigree demonstrating inheritance of a sex-linked trait is the family of Queen Victoria of England and hemophilia.

Queen Victoria had four sons and five daughters. Her son Leopold had hemophilia and died as a result of a minor fall. Two of her daughters, Alice and Beatrice, were carriers for the trait and passed the disorder to royal families in Spain, Prussia, and Russia over four generations.

The Spanish royal family  Victoria’s daughter Beatrice, a carrier for the trait, married Prince Henry of Battenberg, a descendent of Prussian royalty. Two of their sons inherited the trait, both dying before the age of 35. Her daughter, Victoria, was a carrier and married King Alfonso XIII of Spain, thus transmitting the allele to the Spanish royal family. Two of their sons died of hemophilia, also by their early thirties.

The Prussian royal family  Alice, another of Victoria’s daughters, married Louis IV of Hesse, part of the Prussian royal family and related to Prince Henry of Battenberg. One of Alice’s sons, Frederick, died at the age of three from hemophilia. One of her daughters, Irene, continued to pass the trait to the next generation of Prussian royalty by giving it to two of her sons.

The Russian royal family  Irene’s sister and Queen Victoria’s granddaughter, Alix (Alexandra), married Czar Nicholas II of Russia. Four healthy daughters were born, but the first male heir, Alexis, showed signs of bleeding and bruising at only six weeks of age. Having a brother, an uncle, and two cousins who had suffered from the disorder and died at early ages, you can imagine the despair Alix felt for her son and the future heir. In desperation, the family turned to Rasputin, a man who claimed to have healing abilities and used Alexis’ illness for his own political power. The series of events surrounding Alexis and his hemophilia played a role in the downfall of the Russian monarchy.

The British throne today  Queen Elizabeth II, the current English monarch, is descended from Queen Victoria’s eldest son, Edward VII. Because he did not inherit the trait, he could not pass it on to his children. Therefore, the British monarchy today does not carry the recessive allele for hemophilia, at least not inherited from Queen Victoria.

If you were the child of a female carrier for a sex-linked trait such as hemophilia, what would be your chances of carrying the trait?

To find out more about hemophilia, visit the Glencoe Science Web site.

science.glencoe.com
## SUMMARIZE

### Section 12.1

**Mendelian Inheritance of Human Traits**

**Main Ideas**
- A pedigree is a family tree of inheritance.
- Most human genetic disorders are inherited as rare recessive alleles, but a few are inherited as dominant alleles.

**Vocabulary**
- carrier (p. 316)
- fetus (p. 318)
- pedigree (p. 315)

### Section 12.2

**When Heredity Follows Different Rules**

**Main Ideas**
- Alleles can be incompletely dominant or codominant.
- There may be many alleles for one trait or many genes that interact to produce a trait.
- Inheritance patterns of genes located on sex chromosomes are due to differences in the number and kind of sex chromosomes in males and in females.
- The expression of some traits is affected by the internal and external environments of the organism.

**Vocabulary**
- autosome (p. 324)
- codominant alleles (p. 323)
- incomplete dominance (p. 321)
- multiple alleles (p. 323)
- polygenic inheritance (p. 326)
- sex chromosome (p. 324)
- sex-linked trait (p. 325)

### Section 12.3

**Complex Inheritance of Human Traits**

**Main Ideas**
- The majority of human traits are controlled by multiple alleles or by polygenic inheritance. The inheritance patterns of these traits are highly variable.
- Sex-linked traits are determined by inheritance of sex chromosomes. X-linked traits are usually passed from carrier females to their male offspring. Y-linked traits are passed only from male to male.
- Mistakes in meiosis, usually due to nondisjunction, may result in an abnormal number of chromosomes. Autosomes or sex chromosomes can be affected.

**Vocabulary**
- karyotype (p. 335)

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### UNDERSTANDING MAIN IDEAS

1. If a trait is X-linked, males pass the X-linked allele to ________ of their daughters.
   - a. all
   - b. half
   - c. none
   - d. 1/4

2. Stem length demonstrates a range of phenotypes. This is an example of ________.
   - a. autosomal dominant
   - b. autosomal recessive
   - c. sex-linkage
   - d. polygenic inheritance
3. Two parents with normal phenotypes have a daughter with a genetically inherited disorder. This is an example of a(n) ________ trait.
   a. autosomal dominant  
   b. autosomal recessive  
   c. sex-linked  
   d. polygenic
4. Which of the following disorders would be inherited according to the pedigree shown here?
   a. Tay-Sachs disease  
   b. sickle-cell anemia  
   c. cystic fibrosis  
   d. Huntington’s disease
5. Which of the following disorders is likely to be inherited by more males than females?
   a. Huntington’s disease  
   b. Down syndrome  
   c. hemophilia  
   d. cystic fibrosis
6. Infants with PKU cannot break down the amino acid ________.
   a. tyrosine  
   b. lysine  
   c. methionine  
   d. phenylalanine
7. A karyotype reveals ________.
   a. an abnormal number of genes  
   b. an abnormal number of chromosomes  
   c. polygenic traits  
   d. multiple alleles for a trait
8. A mother with blood type I^P_i and a father with blood type I^A_iB^b have children. Which of the following genotypes would be possible for their children?
   a. AB  
   b. O  
   c. B  
   d. a and c are correct
9. Normally, lethal autosomal dominant traits are eliminated from a population because they ________.
   a. have a late onset  
   b. have an early onset  
   c. don’t produce phenotypes that affect a carrier’s health  
   d. aren’t dominant
10. Whose chromosomes determine the sex of offspring in humans?
    a. mother’s  
    b. father’s  
    c. both parents’  
    d. neither parents’
11. A single individual carries ________ alleles for a trait.
12. ________ is a disorder that results from trisomy of chromosome 21.
13. Most sex-linked traits are passed from mother to ________.
14. The normal sex chromosomes of human males are ________, and the normal sex chromosomes of females are ________.
15. To analyze ________, geneticists make a chart of chromosomes called a(n) ________.
16. ________ during meiosis might result in monosomy or trisomy.
17. The inheritance pattern that occurs equally in both sexes and skips generations is ________.
18. The genotype of the individual represented by this pedigree symbol is ________.
   Use the letters Y and y to represent alleles.
19. Feather colors in pigeons are produced by ________ inheritance.
20. If a trait has three different phenotypes, the trait is inherited by ________ or ________.

APPLYING MAIN IDEAS

21. The brother of a woman’s father has hemophilia. Her father was unaffected, but she worries that she may have an affected son. Should she worry? Explain.
22. If a child has type O blood and its mother has type A, could a man with type B be the father? Why couldn't a blood test be used to prove that he is the father?

23. Why do certain human genetic disorders, such as sickle-cell anemia and Tay-Sachs disease, occur more frequently among one ethnic group than another?

24. How can a single gene mutation in a protein such as hemoglobin affect several body systems?

**Thinking Critically**

25. Recognizing Cause and Effect Explain why a male with a recessive X-linked trait usually produces no female offspring with the trait.


27. Concept Mapping Complete the concept map by using the following vocabulary terms: sex-linked trait, autosomes, karyotype, sex chromosomes, polygenic inheritance, codominant alleles.

28. If a child has type O blood and its mother has type A, could a man with type B be the father? Why couldn’t a blood test be used to prove that he is the father?

29. Why do certain human genetic disorders, such as sickle-cell anemia and Tay-Sachs disease, occur more frequently among one ethnic group than another?

30. How can a single gene mutation in a protein such as hemoglobin affect several body systems?

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**Assessing Knowledge & Skills**

The following graph illustrates the number of flowers produced per plant by a certain plant population.

**Interpreting Data** Use the graph to answer the questions that follow.

1. How many flowers are produced by plants that have only dominant genes for flower production?
   - a. 4
   - b. 12
   - c. 16
   - d. 28

2. How many flowers are produced by plants that have half the possible number of dominant genes for flower production?
   - a. 4
   - b. 12
   - c. 16
   - d. 28

3. What pattern of inheritance is suggested by the graph?
   - a. multiple alleles
   - b. incomplete dominance
   - c. polygenic inheritance
   - d. sex-linkage

4. Observing and Inferring From the above graph, estimate the number of gene pairs that control the number of flowers in these plants.

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**CD-ROM**

For additional review, use the assessment options for this chapter found on the *Biology: The Dynamics of Life Interactive CD-ROM* and on the Glencoe Science Web site. science.glencoe.com