CHAPTER
17

Patterns and Processes in Inheritance

Chapter Concepts

17.1 Laying Foundations: Peas, Patterns, and Probabilities

- All somatic cells of diploid organisms have two alleles (copies) of each gene. When a gamete forms, it receives only one of these two alleles.
- The distribution of alleles in gametes is random.
- The inheritance of characteristics follows predictable patterns.

17.2 Extending Mendel's Laws: More Patterns and Probabilities

- Genes are arranged in a linear manner along chromosomes.
- Alleles for genes that are close together on the same chromosome do not assort independently.
- The probability of recombination of linked genes increases with the map distance between these genes.
- Genes that are located on sex chromosomes have a distinct pattern of inheritance.
- The expression of certain genes may be influenced by other genes and by environmental factors.

17.3 Genetics and Society

- Deliberate selection of particular traits can lead to the development of new breeds of plants and animals.
- The pattern of inheritance of human traits is usually studied through the analysis of pedigrees.
- Genetic screening and diagnosis can determine whether an individual carries genes for a particular genetic condition.

Admired for its speed, stamina, and good nature, the Appaloosa is one of the many gifts Aboriginal peoples have bestowed on the world. Horses (*Equus conversidens*) were once native to North America, but they became extinct about 10 000 years ago, likely as a result of environmental change and overhunting. Spanish explorers reintroduced horses to North America in the 1500s. Many First Nations peoples quickly became expert horse handlers. The Nez Perce became, and remain, famous for their selective breeding of horses. Living by the Palouse River near the Rocky Mountains, the Nez Perce carried out a very successful breeding program, selecting only the best and strongest horses for mating. Originally called the Palouse horse, after the Palouse River, this breed soon became known as the Appaloosa.

Launch Lab

Coin Toss

The Nez Perce developed their selective breeding prowess through skills such as observation and patience. Modern geneticists use these same skills and others, as well as knowledge from biochemistry, statistical analysis, and other fields of inquiry. Geneticists analyze the data they collect, and they may use the results to formulate or to test a hypothesis.

How well can you predict results based on a hypothesis? How close to the predicted results must the data be for you to be confident that they support the hypothesis? A coin toss is a good way to make and test predictions. You and a partner will each toss a coin. Then you will record the results to show whether or not either of the two coins has turned up heads. Either two heads or a combination of a head and a tail will be considered "heads." Two tails will be considered "no heads." You will repeat the coin toss 10 times.

Materials

2 coins

Procedure

1. Determine the probability that any toss of two coins will result in "heads" or "no heads." Use a table, such as the one below, to determine the probability. For 10 pairs of coin tosses, how many "heads" do you expect? How many "no heads" do you expect?
2. Toss your coins 10 times, and record your results.

Results	Prediction "Heads" (Head–Head or Head–No head)	Prediction "No Heads" (No head–No head)	Actual "Heads" (Head–Head or Head–No head)	Actual "No Heads" (No head–No head)
Toss Results				
Total				

Analysis

1. Calculate your percent error in the number of "heads" by using the formula below.

$$\text{percent error} = \left|\frac{\text{observed} - \text{expected}}{\text{expected}}\right| \times 100\%$$

2. Do your data support your hypothesis? Why or why not?
3. Combine the data obtained by the entire class. Calculate the percent error in the number of "heads." How does the percent error for the combined data compare with yours?

The genetic information that is responsible for eye colour only codes for the presence or absence of a pigment called melanin. Melanin produces either brown or blue colouring. What could be responsible for other eye colours in humans?

SECTION 17.1

Laying Foundations: Peas, Patterns, and Probabilities

Section Outcomes

In this section, you will

- **describe** the evidence for segregation and the independent assortment of alleles
- **compare** ratios and probabilities of genotypes and phenotypes for dominant and recessive alleles, incompletely dominant alleles, and co-dominant alleles
- **perform** an experiment to demonstrate the inheritance of a trait controlled by a single gene
- **interpret** patterns and trends in data from monohybrid and dihybrid inheritance

Key Terms

selective breeding
true breeding
monohybrid cross
dominant
recessive
complete dominance
law of segregation
genotype
phenotype
homozygous
heterozygous
Punnett square
test cross
dihybrid cross
law of independent assortment
incomplete dominance
co-dominance
chromosome theory of inheritance

Eurasian wolf (*Canis lupus lupus*) | Great Pyrenees (*Canis familiaris*) | Dachshund (*Canis familiaris*) | Toy poodle (*Canis familiaris*)

Figure 17.1 The Great Pyrenees is one of the oldest-known dog breeds, bred several thousand years ago to protect sheep herds from wolves and bears. The dachshund was bred about 600 years ago to hunt badgers in their underground dens. The toy poodle, a product of the mid-twentieth century, was bred to provide dog lovers with the intelligence and sensitivity of the large poodle in a much smaller form.

For thousands of years, humans have recognized that offspring resemble their parents. People have used this observation to their advantage by choosing and breeding specific plants and animals for particular physical features or behaviours—a process called **selective breeding**. Beginning with wild canines, such as the Eurasian wolf (*Canis lupus lupus*), humans have used centuries of selective breeding to develop, gradually, hundreds of breeds of dogs with specific attributes (see Figure 17.1).

Early Theories of Inheritance

People bred animals and plants for thousands of years without understanding the mechanisms of inheritance. Many people were curious about these mechanisms, however, and tried to explain them. The Greek philosopher Aristotle (384–322 B.C.E.) proposed the first widely accepted theory of inheritance, called *pangenesis*. According to this theory, egg and sperm consist of particles, called pangenes, from all parts of the body. Upon fertilization of the egg by a sperm, the pangenes develop into the parts of the body from which they were derived. The theory of pangenesis was accepted for hundreds of years although no experiments were done to test its assumptions or results.

In 1677, the amateur scientist Antony van Leeuwenhoek (1632–1723) discovered living sperm in semen with his exquisitely designed single-lens microscopes. He believed that he saw a complete, miniature person, called a homunculus, in the head of sperm (see Figure 17.2). Leeuwenhoek believed that the homunculus came from the father but developed in the mother. A contemporary of Leeuwenhoek, Regnier de Graaf, proposed that the egg, not the sperm, contained the entire person. He argued that the sperm only stimulated the egg to develop.

During the 1800s, when the breeding of ornamental plants was becoming popular, scientists observed that the offspring had characteristics of *both* parent plants. The idea of blending became the working theory of inheritance. Scientists believed that characteristics of the parents blended in the offspring in a way that was irreversible. In other words, scientists

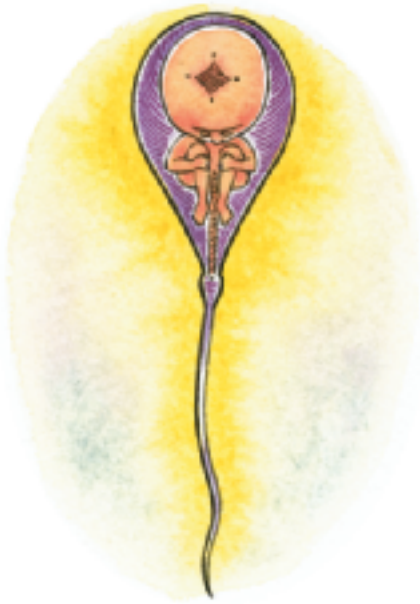


Figure 17.2 Even with microscopes as powerful as those made by Antony van Leeuwenhoek, able to magnify up to 500 times, details of the human sperm cell left much to the imagination of the observer.

believed that the original parental characteristics would not reappear in future generations.

None of the explanations of inheritance proposed prior to the 1850s stood the test of time.

Q1 What is selective breeding? Give an example.

Q2 Briefly describe two early explanations of inheritance.

Developing a Theory of Inheritance: Gregor Mendel's Experiments

Great scientific discoveries are sometimes made in the most unexpected places. No one would have expected a great discovery to be made in a monastery garden by an Augustinian monk in Brunn, Austria (now in the Czech Republic), in the 1860s. The scientists of the time certainly did not expect it. In fact, they ignored the scientific paper describing this discovery until 1900, when supporting evidence began to emerge. Nevertheless, the work of a monk and teacher named Gregor Mendel (1822–1884) laid the foundation for the field of genetics, the science of inheritance (see Figure 17.3).

Between the years 1856 and 1863, Mendel bred, tended, and analyzed over 28 000 pea (*Pisum sativum*) plants in the monastery garden. He observed many different traits, or characteristics. He chose seven traits that were expressed in two easily distinguishable forms. These traits are shown in Figure 17.4. Before

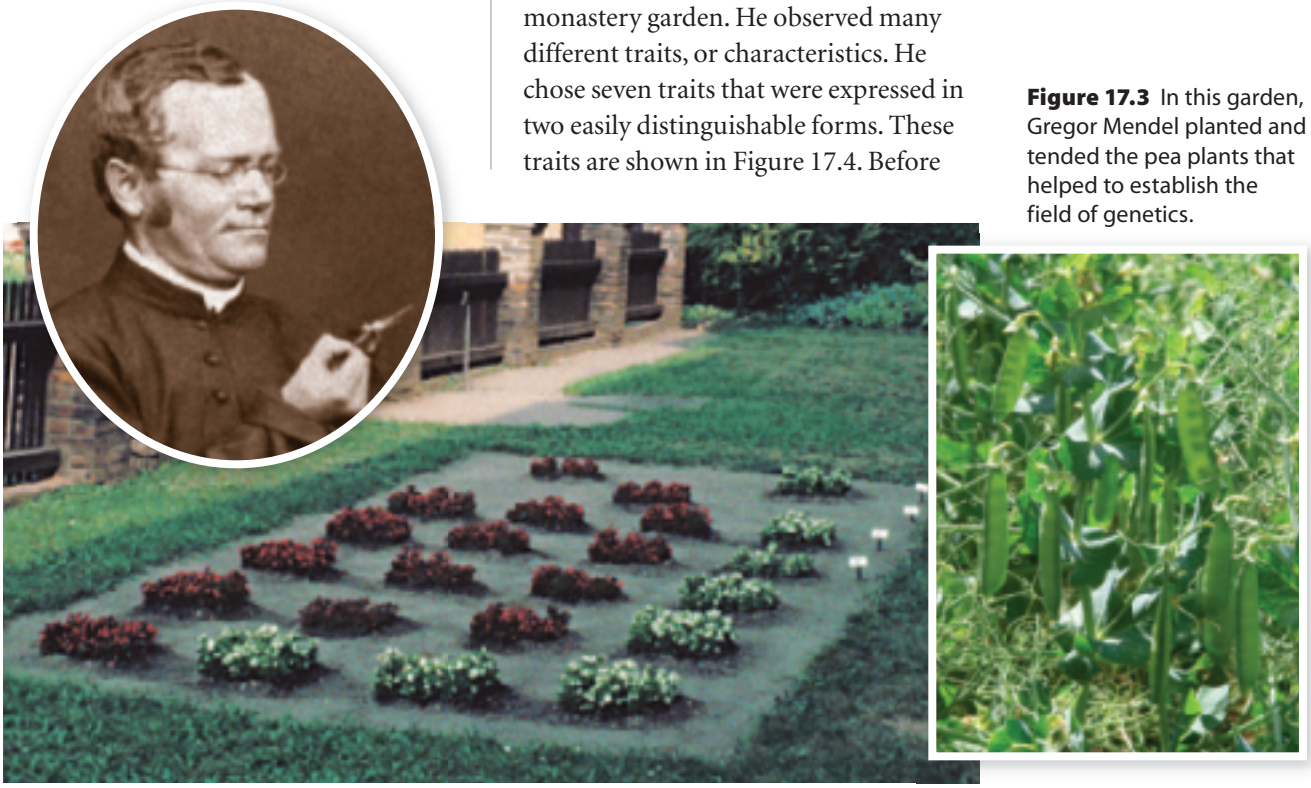
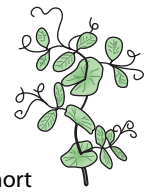
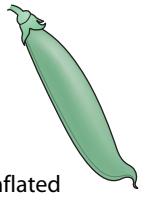
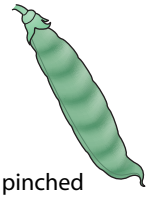
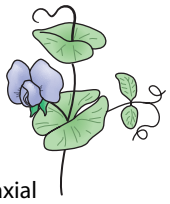
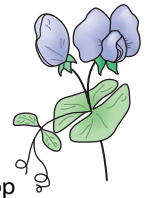
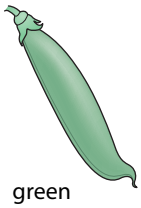
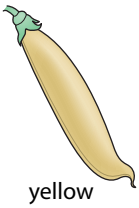


Figure 17.3 In this garden, Gregor Mendel planted and tended the pea plants that helped to establish the field of genetics.

doing any experiments, Mendel let the plants self-pollinate to ensure that they were true breeding. **True breeding** plants exhibit the same characteristics generation after generation.

Mendel called the true breeding plants the *parental*, or P, generation. He started his experiments by crossing true breeding plants for each characteristic with true breeding plants having the opposite characteristic. For example, he bred plants that produced purple flowers with plants that produced white flowers. He called the offspring the first *filial*, or F_1, generation. Mendel let the F_1 plants self-pollinate and then observed the characteristics of their offspring, which he called the second filial, or F_2, generation. Mendel identified the characteristics of the F_1 and F_2 generation plants and counted how many plants produced which characteristics. Since only one trait is involved in this type of cross, it is called a **monohybrid cross**. (A *hybrid* is the offspring of a cross between two parent organisms with different inheritable traits.)

Figure 17.4 Each of the seven traits that Mendel chose to study had two clearly distinguishable forms.

Characteristics	Contrasting traits	
stem length	 tall	 short
pod shape	 inflated	 pinched
seed shape	 round	 wrinkled
seed colour	 yellow	 green
flower position	 axial	 top
flower colour	 purple	 white
pod colour	 green	 yellow

Q3 Explain how a hybrid plant is different from a true breeding plant.

Q4 Explain the difference between P, F_1, and F_2 generations in a cross.

Dominant and Recessive Genes

Mendel observed that, for every trait, the F_1 plants showed only one of the two parental characteristics. For example, in the cross between plants with round seeds and plants with wrinkled seeds, all the seeds in the F_1 generation were round. Although all the F_1 plants had a copy of each form of the factor for seed shape, only one form was shown, or *expressed*. Mendel called the characteristic that was expressed in the F_1 generation **dominant**. He called the characteristic that was not expressed **recessive**. Mendel concluded that one form showed **complete dominance** over the other form. That is, an individual with one recessive and one dominant form had the same observable physical characteristic as an individual with two dominant forms.

Table 17.1 summarizes the dominant and recessive forms of each of the seven traits that Mendel tested. If you compare Table 17.1 with Figure 17.4, you will see that the forms on the left side of the second column are dominant and the forms on the right are recessive.

The Law of Segregation

When Mendel analyzed the data from his monohybrid crosses, he saw very clear patterns. For each of the seven traits he analyzed, each F_1 plant exhibited the trait of only one of the two parental plants. For example, when he crossed plants that produced round seeds with plants that produced wrinkled seeds, the F_1 plants all produced round seeds. When the F_1 plants self-pollinated, however, some of their offspring (the F_2 generation) had wrinkled seeds.

In repeated experiments, Mendel found that traits that had not appeared in the F_1 plants reappeared in the F_2 plants, but in smaller numbers than in the P generation. Figure 17.5 summarizes the results of one of Mendel's experiments, in which he crossed parental plants with different seed shapes. Notice that the ratio of F_2 plants with round seeds to F_2 plants with wrinkled seeds is about 3:1. Mendel observed similar ratios in the F_2 generation for monohybrid crosses involving each of the seven traits.

From these data for monohybrid crosses, Mendel inferred the following generalizations:

- Discrete factors determine individual traits. (**Note:** Mendel used the term "factors" to describe what are now called genes.)
- Each individual organism has two copies of each factor.
- When gametes (eggs and sperm) are formed, the copies of the factors segregate so that each gamete receives one copy of each factor.
- Eggs and sperm fuse randomly. The embryo that develops into a new individual has two copies of each factor—one copy from each parent.

Mendel's first law, also called the **law of segregation**, is a summary of these concepts.

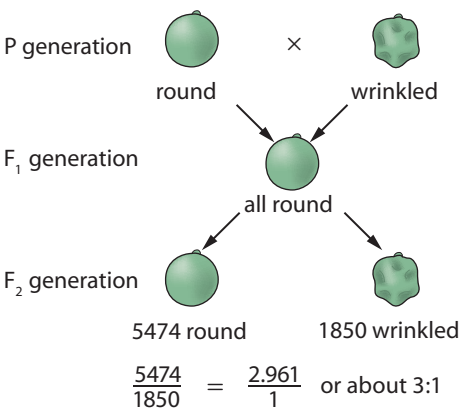


Figure 17.5 The ratio of plants with round seeds to plants with wrinkled seeds in the F_2 generation is 5474:1850 or 2.96:1. This is very close to a 3:1 ratio.

Mendel's First Law: The Law of Segregation

All individuals have two copies of each factor. These copies segregate (separate) randomly during gamete formation, and each gamete receives one copy of every factor.

Q5 State Mendel's first law in your own words, and use an example to illustrate it.

Representing Genetic Crosses

In 1909, Danish botanist and geneticist, Wilhelm Ludwig Johannsen, coined the term gene for Mendel's factors. The different forms of each gene are now called alleles. That is, you would say that the *gene* is seed shape, and the *allele* for round seeds is dominant to the *allele* for wrinkled seeds (or the *allele* for wrinkled seeds is recessive to the *allele* for round seeds). To symbolize the different alleles,

Table 17.1 The Seven Traits of Pea Plants Studied by Mendel

Trait	Dominant	Recessive
stem length	tall	short
pod shape	inflated	pinched
seed shape	round	wrinkled
seed colour	yellow	green
flower position	axial	terminal
flower colour	purple	white
pod colour	green	yellow

BiologyFile

FYI

Before Mendel did his experiments, heredity had been a descriptive science. No one had ever applied mathematical methods or statistical analysis to the study of inheritance.

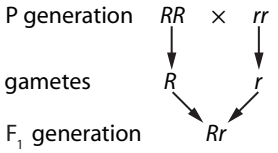


Figure 17.6 Representation of a monohybrid cross between two true breeding parent plants. One parent plant is homozygous for round seeds (*RR*), and the other parent plant is homozygous for wrinkled seeds (*rr*). The F_1 generation is heterozygous for round seeds (*Rr*).

geneticists use a system that Mendel devised. They use the first letter in the description of the dominant allele, in upper case, to represent the dominant allele. They use the *same* letter in lower case to represent the recessive allele.

According to this system, the allele for round seeds is represented by *R* and the allele for wrinkled seeds is represented by *r*. Since each individual has two alleles for every gene, a pea plant could have the allele combination *RR*, *Rr*, or *rr* (see Figure 17.6). Johannsen called the combination of alleles for any given trait the individual's **genotype**. He called the outward expression of the trait—the physical form that you can observe—the **phenotype**. For example, an individual with genotype *RR* would have the phenotype of round seeds. Since *R* is dominant to *r*, individuals with genotype *Rr* would also have the phenotype of round seeds. Only individuals with genotype *rr* would have wrinkled seeds.

An individual with two identical alleles for a trait, such as *RR* or *rr*, is **homozygous** for the trait. An individual with two different alleles for a trait, such as *Rr*, is **heterozygous** for the trait.

You can use these symbols and concepts to represent Mendel's crosses of plants with round and wrinkled seeds in a condensed form. Since the parental plants were true breeding, they had to have identical alleles. You would write the cross as shown in Figure 17.6.

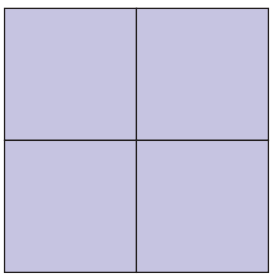
6. State the meanings of the terms dominant and recessive.
7. Distinguish between the terms gene and allele.
8. Distinguish, using an example, between the terms genotype and phenotype.
9. What does it mean to be homozygous for a trait? How is this different from being heterozygous for a trait?

Analyzing Genetic Crosses

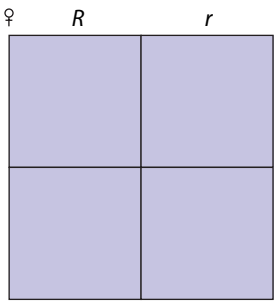
How did the recessive characteristic reappear in the F_2 generation? When F_1 plants produce gametes, the alleles segregate randomly and each gamete receives only one allele. There is an equal chance that any gamete will receive *R* or *r*. To analyze the results of crosses, British geneticist Reginald Punnett (1875–1967)

Figure 17.7 Using a Punnett square to analyze a cross between F_1 plants, producing an F_2 generation assessed for seed shape

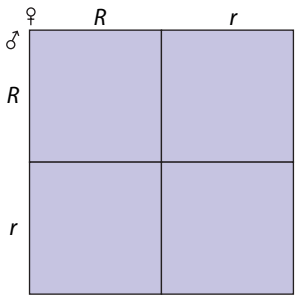
A When working with one gene, make a box and divide it into four squares.



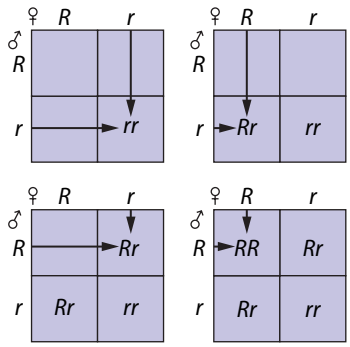
B Above the squares, write the genotypes of the gametes from the female parent. The ♀ symbol represents gametes from the female parent. (Some resources do not include this symbol.)



C Beside the squares, on the left, write the genotypes of the gametes from the male parent. The ♂ symbol represents gametes from the male parent.



D Inside each square, write the symbols for the allele above it and the allele beside it. The genotypes inside the squares are the genotypes of the offspring in the ratio that would be expected for the cross.



devised a visual technique that is now called the **Punnett square**. This technique, applied to the F_1 cross, is shown in Figure 17.7.

After drawing a Punnett square, you can analyze the genotypes to determine the phenotypes. In Figure 17.7, for example, all the *RR* and *Rr* plants will produce round seeds, and all the *rr* plants will produce wrinkled seeds. The ratio of the phenotypes, round to wrinkled, is 3:1. This is the ratio that Mendel found in his data. As noted in Figure 17.8, Mendel worked backward to formulate a hypothesis that could explain the data. His law of segregation explains the data very well. In the Practice Problems below, you will see this for yourself. Then, in the investigation that follows, you will assess the law of segregation.

Test Cross

When geneticists want to know if a phenotypically dominant individual is homozygous or heterozygous, they do a test cross. A **test cross** is a cross between the organism of unknown genotype and a homozygous recessive organism. In mice, for example, a condition called waltzer is recessive. A waltzer mouse has a defect in the region of the inner ear that interferes with its balance. Consequently, waltzers run in circles. A mouse that runs normally might be homozygous dominant for this gene, or it might be heterozygous. If geneticists had several mice that walk normally, and wanted to know if any of these mice were heterozygous for the waltzer mutation, they would do a test cross.

You can use Punnett squares to predict the genotypes and phenotypes of

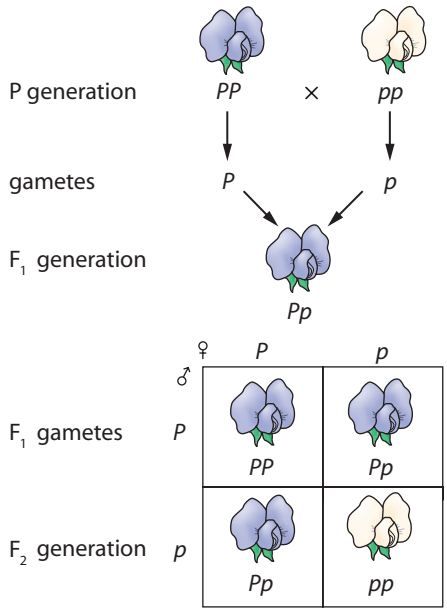


Figure 17.8 For each trait that he tested, Mendel observed the same types of results and inferred the same pattern. This illustration shows a cross between true breeding purple-flowered plants and true breeding white-flowered plants. The ratio of phenotypes in the F_2 generation is 3:1.

the offspring of the test crosses as shown in Figure 17.9. In these examples, *N* represents normal (non-waltzer), and *n* represents the recessive waltzer mutation. The genotypes of the gametes for homozygous recessive are on the top of each Punnett square. One of the two possible genotypes of the mouse being tested is on the side of each Punnett square.

If the mouse being tested is homozygous normal, all the offspring will be phenotypically normal. If the mouse being tested is heterozygous for waltzer, half the offspring will be phenotypically normal and half will be waltzers. As you can see, test crosses are an important tool for geneticists, particularly in cases where reproduction is frequent and multiple offspring are produced.

The Law of Independent Assortment

Mendel did more than conduct experiments to test single traits. He

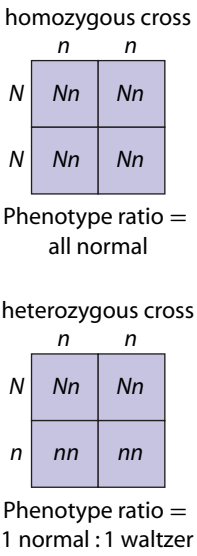


Figure 17.9 Predicting phenotypes of a test cross

Practice Problems

1. Mendel crossed true breeding plants that had yellow pods with true breeding plants that had green pods. Using the information in Table 17.1, predict and write down the genotypes and phenotypes of the F_1 and F_2 generations. Predict the ratio of F_2 plants with green pods to F_2 plants with yellow pods.

2. In one of his experiments, Mendel counted 6022 yellow seeds and 2001 green seeds. Write the genotypes and phenotypes of the plants in all the crosses he did in order to get these results. How well did his data fit the predicted ratios?

INVESTIGATION 17.A

Target Skills

Performing an experiment to demonstrate inheritance of a trait that is controlled by a single pair of genes

Using a Punnett square to interpret patterns and trends in data

Testing the Law of Segregation

In this investigation, you will work with plants called Wisconsin Fast Plants™ (*Brassica rapa*). They germinate and mature quickly, so you can grow your own plants, tend them, pollinate the flowers, and harvest and plant the seeds. Your goal is to collect evidence to explain how stem colour in *Brassica rapa* is inherited. You will use your evidence to predict the phenotype of the male parental generation (P_M). The trait you are focussing on, stem colour, is controlled by a gene that regulates whether or not anthocyanin (a purple pigment) is expressed.

Hypothesis

Each group will formulate a testable hypothesis to explain how the stem colour phenotypes of *Brassica rapa* reflect their genotypes. Your hypothesis will form the basis of your experimental design.

Safety Precautions

Wash your hands whenever you handle any of the materials in this Investigation.

Materials

- *Brassica rapa* seeds and growing systems
- potting mix
- fertilizer
- stakes and ties
- labels
- instructions for growing, tending, pollinating, and harvesting

Procedure

1. One group will plant seeds from the female parental generation (P_F). All the other groups will plant seeds from the first-generation offspring (F_1). Use the instructions provided by your teacher to germinate the seeds and tend the developing plants.
2. After about four days, observe the stem colour and all other phenotypes of the young P_F and F_1 plants. Record your observations in a suitable table.
3. Return the P_F plants to your teacher. Continue to tend the F_1 plants by thinning the plants to two per pot. Refer to the instructions provided by your teacher.
4. In your group, discuss how you think stem colour is inherited in *Brassica rapa*. State a hypothesis and use it to predict the stem colour of the male parental generation plants (P_M) from which the F_1 offspring came.
5. After about a week and a half, pollinate the F_1 plants over three days. Be sure that all the flowers receive pollen from several different plants. (Refer to the instructions provided by your teacher.)
6. Based on your hypothesis, predict the stem colour of the second-generation offspring (F_2) that will result from the pollination you have done.
7. Over the next several days, cut off any new flower buds that developed after you pollinated the plants.
8. After about two weeks, stop watering the plants. Let them dry for a full week.
9. After about one week, harvest the seeds from the pods of the F_1 plants. (Refer to the instructions.) These are your seeds for the F_2 generation. Plant the seeds.
10. After about four days, observe the stem colour and other phenotypes of the young F_2 plants. Design a table to record your observations.
11. Return the F_2 plants to your teacher.
12. After you complete Analysis questions 1 and 2, plant the P_M seeds supplied by your teacher. About four days later, record your observations of stem colour and other phenotypes of the young P_M plants in a suitable table.
13. Return all the plants and other materials to your teacher.

Analysis

1. Think about what you know of Mendel and his experimental procedures. In what ways are your experiences similar? In what ways are they different?
2. Explain the purpose of cutting off new flower buds in step 7 of the Procedure.

Conclusions

3. Examine your hypothesis and your results. How accurate was your hypothesis? Identify any sources of error.
4. Draw a Punnett square and a diagram showing the P, F_1, and F_2 generations to record the results of your investigation.

also conducted experiments to find out whether the pattern of segregation of the alleles for one gene has any influence on the pattern of segregation of the alleles for another gene. He crossed plants that were true breeding for two different traits with plants that were true breeding for the opposite form of the same two traits. Since two genes are involved in this type of cross, it is called a **dihybrid cross**.

In one dihybid cross, for example, Mendel crossed true breeding tall plants that had green pods (*TTGG*) with true breeding short plants that had yellow pods (*ttgg*). This produced an F_1 generation of plants that were all heterozygous for both traits (*TtGg*). Mendel allowed the F_1 plants to self-pollinate and then analyzed the traits of the F_2 plants.

The cross *TtGg* × *TtGg* produced F_2 plants with the phenotypes of tall with green pods, tall with yellow pods, short with green pods, and short with yellow pods in a ratio of 9:3:3:1. For every dihybrid cross that Mendel carried out and analyzed, he found the same pattern in the F_2 generation. Mendel did a statistical analysis to compare this 9:3:3:1 ratio to the ratio that would be expected if the segregation of alleles for one gene had *no influence* on the segregation of alleles for another gene.

You can use a Punnett square to test Mendel's ratio (see Figure 17.10). All the individuals in the F_1 generation receive one allele for each gene from each parent. The genotype of all F_1 plants is therefore *TtGg*. If the alleles for the two genes segregate independently of each other, the gametes will have an equal chance of carrying any one of the four genotypes, *TG*, *Tg*, *tG*, *tg*. You can use these gametes to construct a Punnett square. As shown in Figure 17.10, the ratio of 9:3:3:1 is obtained when the alleles segregate randomly among the gametes. Mendel based his second law, the **law of independent assortment**, on results such as these.

Mendel's Second Law: The Law of Independent Assortment

The two alleles for one gene segregate (assort) independently of the alleles for other genes during gamete formation.

10. What is a Punnett square?
11. What is a test cross?
12. State Mendel's second law in your own words.

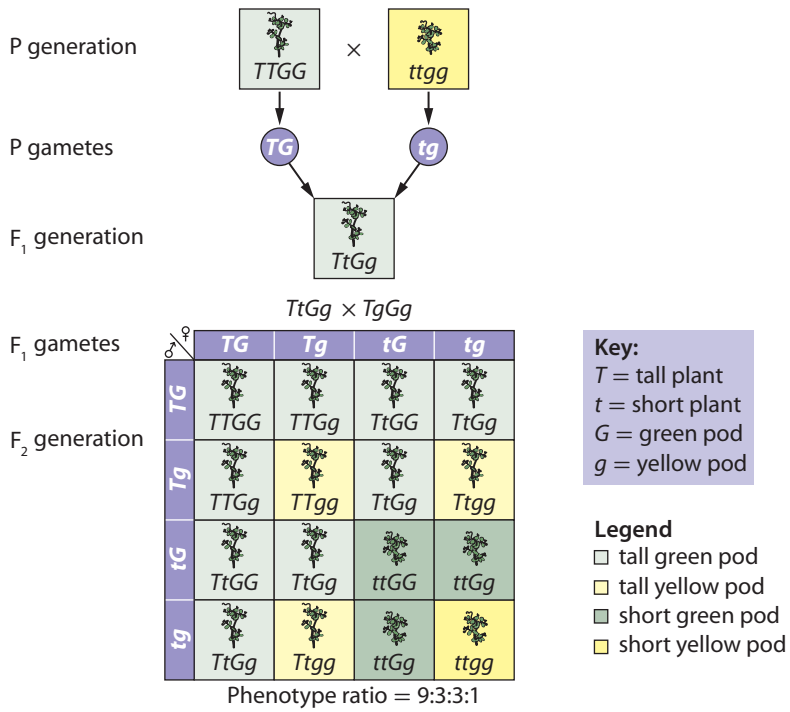


Figure 17.10 For any dihybrid cross, individuals in the largest group (9) have at least one dominant allele for each gene (*T_G_*), where the underscore represents any one of the four alleles. In the intermediate groups (3), the individuals have at least one dominant allele for one gene but two recessive alleles for the other gene (*T_gg* or *ttG_*). The smallest group (1) is homozygous recessive for both traits (*ttgg*).

Incomplete Dominance and Co-dominance

Since Mendel's work became recognized, geneticists have studied a number of traits that do not appear to follow the same pattern of inheritance that Mendel observed. These patterns can also be explained in terms of Mendel's laws, however.

Incomplete dominance describes a condition in which neither of two alleles for the same gene can completely conceal the presence of the other. One example is the flower colour of the four-o'clock plant (*Mirabilis jalapa*). As you can see in Figure 17.11, a cross between a true-breeding red-flowered plant and a true-breeding white-flowered plant produces offspring with pink flowers. When representing incomplete dominance, upper-case and lower-case letters are not generally used to represent the alleles. Some geneticists use all upper-case letters, with subscripts to denote the alleles, as shown in the key in Figure 17.11.

The fact that the alleles segregate just like the alleles for a completely dominant trait is supported by the pattern observed when the pink flowers are self-pollinated. The Punnett square in Figure 17.11 shows that the ratio of red (R_1R_1) to pink (R_1R_2) to white (R_2R_2) flowers should be 1:2:1. The experimentally observed ratios are close enough to the expected ratio of 1:2:1 to support the theory of incomplete dominance.

Two genetic conditions in humans exhibit incomplete dominance: sickle cell anemia and familial hypercholesterolemia. Sickle cell anemia is caused by a specific form of the gene that directs the synthesis of hemoglobin, the molecule that carries oxygen in the blood. Slightly lowered oxygen concentrations in the blood cause the hemoglobin in the red blood cells to form needle-like crystals that distort the shape of the cells. The misshapen cells cannot pass through small capillaries and cause blockages that result in tissue damage to the local area as well as intense pain. Geneticists believe that this form of the gene arose in Africa centuries ago.

The allele for normal hemoglobin is represented as Hb^A, and the allele for sickle cell hemoglobin is represented as Hb^S. As shown in Figure 17.12, individuals who are homozygous (Hb^SHb^S) have sickle cell disease.

Individuals who are heterozygous for the gene (Hb^AHb^S) are said to have the sickle cell trait. Their blood cells are much less likely to become sickle-shaped, and they rarely experience any symptoms. In fact, having the sickle cell trait can be an advantage because heterozygotes are resistant to malaria. This is very beneficial in certain parts of Africa, where deadly malaria epidemics can occur. The sickle cell trait is an example of the principle of *heterozygote advantage*—heterozygous individuals having an advantage over homozygous dominant or homozygous recessive individuals.

Familial hypercholesterolemia is a genetic condition that prevents the tissues from removing low-density lipoproteins (LDL, commonly known

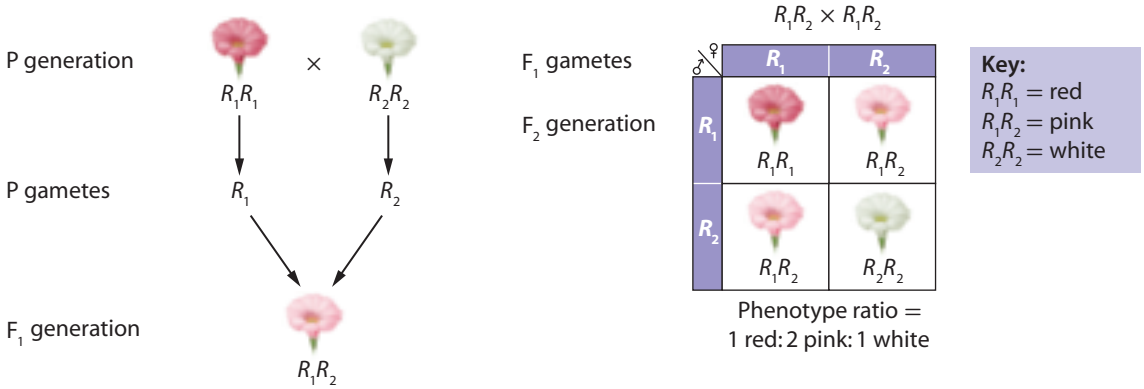


Figure 17.11 The allele for red flowers in the four o'clock plant directs the synthesis of red pigment. When only one allele is present, the flower cannot make enough pigment to make the flowers red, resulting in incomplete dominance (pink flowers).

as "bad cholesterol") from the blood. People who are homozygotes for the trait have six times the normal amount of cholesterol in their blood and may have a heart attack by the age of 2. Heterozygotes have about twice as much cholesterol in their blood and may have a heart attack by the age of 35.

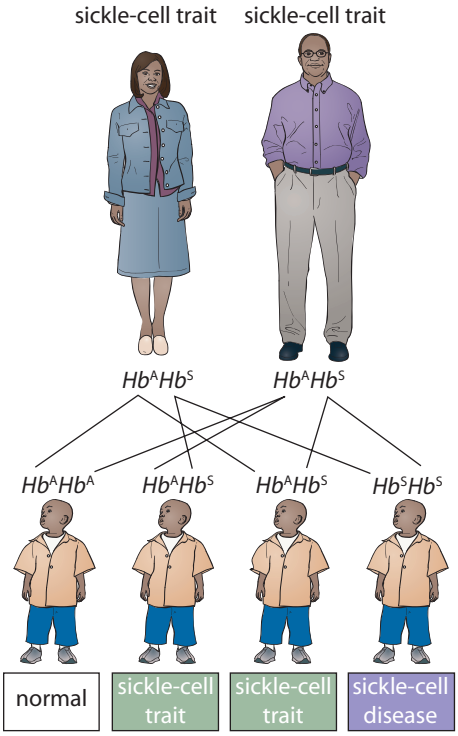
Co-dominance is a situation in which both alleles are fully expressed. A roan horse or cow is an excellent, visible example of co-dominance. A roan animal is a heterozygote in which both the base colour and white are fully expressed. If you look closely at the individual hairs on a blue roan, such as the horse in Figure 17.13, you will see a mixture of black hairs and white hairs. One allele is expressed in the white hairs, and the other allele is expressed in the black hairs. A red roan has a mixture of chestnut-coloured hairs and white hairs.

13 Explain what incomplete dominance is.

14 Give two examples of genetic conditions in humans that exhibit incomplete dominance, and briefly describe what they are.

15 Explain how co-dominance is different from incomplete dominance.

16 Explain why the roan colouring of a horse is an example of co-dominance.



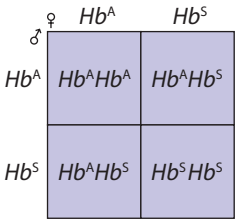


Figure 17.12 When a man and a woman are both heterozygous for the sickle cell gene, there is a one in four chance that they will have a child with sickle cell disease.

Genes and Chromosomes

Throughout his research, Mendel made no attempt to determine the chemical or physical nature of his factors (now called genes). He accepted that they must be present in order to cause the results he observed. This way of thinking was uncommon for the biologists of his time, and it is one reason why his published work went almost unnoticed for many years. It was not until about 1900, about 16 years after Mendel's death, that other biologists began to perform experiments similar to Mendel's experiments with garden peas. Three scientists, working independently, rediscovered Mendel's

Figure 17.13 The roan colouring of a horse usually does not affect the head, mane, and tail. This horse's body looks blue because black and white hairs are thoroughly mixed.

publication. Dutch plant physiologist Hugo de Vries (1848–1935), German botanist Carl Erich Correns (1864–1933), and Austrian agronomist Tschermak von Seysenegg (1871–1962) were all performing crosses with plants. As part of their research, each of them searched the scientific literature for any similar work that had been published. They all found Mendel's publication, and they all realized that their own work was in agreement with Mendel's proposed laws.

Then, in 1902, Walter Sutton (1877–1916), a graduate student at Columbia University in New York, studied sperm development in grasshoppers. Sutton examined the processes of synapsis (segregation of homologous chromosomes) and migration of sister chromatids during meiosis I and meiosis II. Sutton realized that the distribution of chromosomes into developing gametes follows the pattern for two alleles of a gene, according to Mendel's law of segregation. Genes come in pairs, as do chromosomes. During gamete formation, alleles segregate just as homologous chromosomes do (see Figure 17.14). Sutton published a paper proposing that genes are carried on chromosomes. Sutton's theory became known as the **chromosome theory of inheritance**.

As you saw in Chapter 16, there is no apparent interaction between non-homologous chromosomes during meiosis. The movement of each pair of homologous chromosomes is independent of the movement of all the other pairs of homologous chromosomes. This agrees with Mendel's law of independent assortment; however, Sutton recognized an important implication of his research: genes that are carried on the same chromosome do not assort independently. This means that they do not follow Mendelian inheritance patterns.

Q17 Give two examples of how the movement of alleles is consistent with the movement of chromosomes during meiosis.

Objections to Sutton's Theory

Sutton's theory—that Mendel's "factors" are located on chromosomes—was not widely accepted at first. Many scientists

Practice Problems

3. In zucchini (*Cucurbita pepo*), yellow-coloured flesh is recessive to white-coloured flesh. You are plant breeder and would like to know if any of the white-fleshed zucchini you have are heterozygous for the yellow-fleshed allele. How would you determine this? Describe the procedure you would follow.

4. In tomatoes (*Lycopersicon esculentum*), red fruit (*R*) is dominant to yellow fruit (*r*), and tall (*T*) is dominant to short (*t*). True-breeding tall plants that produced red fruit were crossed with true-breeding short plants that produced yellow fruit.

a) State the genotype and phenotype of the F_1 generation plants.

b) List the genotypes of the gametes produced by the F_1 plants.

c) List the genotypes and phenotypes of the F_2 generation plants. Include the genotypic and phenotypic ratios of the F_2 generation plants.

5. In mice, black fur (*B*) is dominant to brown fur (*b*), and non-waltzer mice (*N*) are dominant to waltzers (*n*). Two mice that are heterozygous for both traits are crossed. Draw a Punnett square for the F_1 generation, and determine the phenotype ratio for these offspring.

6. The gene that codes for colour in snapdragons (*Antirrhinum majus*), exhibits incomplete dominance. A true-breeding red snapdragon is crossed with a true-breeding white snapdragon. What is the phenotype ratio of the F_1 generation? The F_1 offspring are then crossed to produce an F_2 generation. Draw a Punnett square for this generation and determine the phenotypic ratio.

7. Two blue roan horses are bred together. What is the chance that the colt will be white?

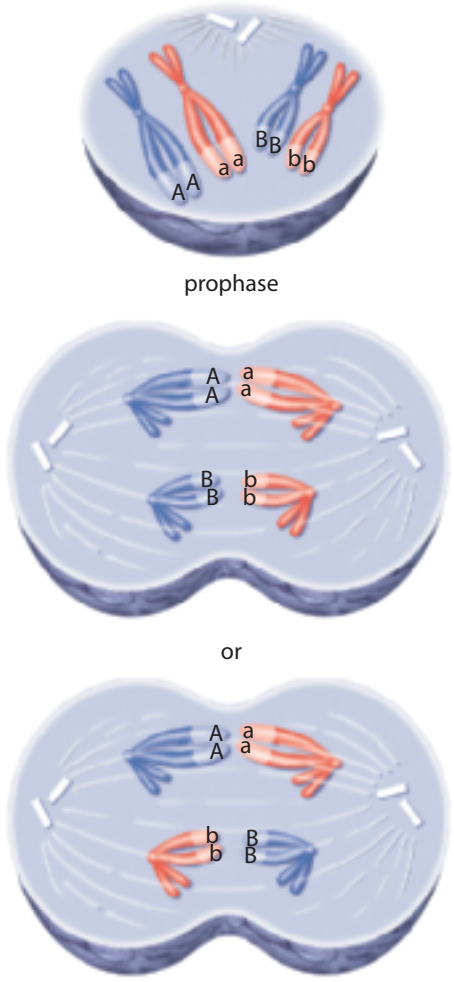


Figure 17.14 Alleles and chromosomes both segregate during meiosis. During anaphase I, the homologous chromosomes segregate (separate) and migrate to opposite ends of the cell. After telophase I, the homologous chromosomes are in separate cells. The resulting gametes are equally likely to contain each possible combination of alleles.

still believed in the blending of hereditary information. Many also believed that the environment, or events in the parents' lives, played the main role in determining which parental traits would be expressed in the offspring. Objections to Sutton's theory included the lack of convincing evidence linking Mendel's factors to any cellular structure and the absence of any studies that could account for more complex inheritance patterns. The path to the resolution of these objections, and to the affirmation of a chromosomal theory of heredity, leads back to Columbia University, to a laboratory room buzzing with intellectual and experimental excitement—and with fruit flies, *Drosophila melanogaster*. You will learn about the important work that occurred in this laboratory in the next section.

Section 17.1 Summary

- Gregor Mendel used pea plant crosses to follow the transmissions of one or two traits at a time. The genes for the traits he studied were carried on different chromosomes, and each gene had two alleles.
- Mendel's law of segregation states that inherited factors (genes) separate in meiosis. Each individual receives one copy of each gene from each parent.
- An allele whose expression masks another is dominant. An allele whose expression is masked by a dominant allele is recessive.
- An individual who has two identical alleles for a trait (for example, *rr*) is homozygous for that trait. An individual who has two different alleles for a trait (for example, *Rr*) is heterozygous for the trait. A heterozygote has two different alleles of a gene. A homozygous recessive individual has two recessive alleles. A homozygous dominant individual has two dominant alleles.
- The combination of alleles is the genotype. The expression of a genotype of an individual is its phenotype.
- The parental generation is designated P_1. The next general is the first filial generation and is designated F_1. The next generation is the second filial generation and is designated F_2.
- A monohybrid cross yields a phenotype ratio of 3:1. A test cross breeds an individual of unknown genotype to a homozygous recessive individual.
- Punnett squares are based on the principles of probability and can be used to predict the outcomes of genetic crosses.
- Mendel's law of independent assortment was derived by observing the transmission of two or more characters whose genes are on different

chromosomes. Because maternally and paternally derived chromosomes (and their genes) assort randomly in meiosis, different gametes receive different combinations of genes.
- A dihybrid cross yields a 9:3:3:1 phenotype ratio.
- Different dominance relationships influence phenotype ratios. Heterozygotes of incompletely dominant alleles have phenotypes intermediate between those of the two homozygotes. Co-dominant alleles are both expressed.
- The chromosome theory of inheritance states that genes are located on chromosomes, and chromosomes provide the basis for the segregation and independent assortment of genes.

Section 17.1 Review

1. Explain how the Launch Lab: Coin Toss applies to making predictions about the genotypes and phenotypes of the F_2 generation of a monohybrid cross.

2. Describe two aspects of Mendel's experiments that were unusual for biology experiments in the 1800s.

3. Compare and contrast the following terms: selective breeding and true breeding. How does each term relate to the field of genetics?

4. Explain the meaning of the law of segregation.

5. If you did not know whether green or yellow was dominant for the gene for pod colour in peas, how would you determine this experimentally? Describe your experiment in detail, and explain how you would interpret the results.

6. In the garden pea, inflated pod shape (*I*) is dominant to constricted shape (*i*). Axial flower position (*A*) is dominant to terminal flower position (*a*). Start with the cross *IIAA* × *iiaa* from the parental generation.

a) State the genotype and phenotype of the F_1 plants.

b) List all of the genotypes and phenotypes of the F_2 plants, and determine the genotypic and phenotypic ratios.

c) Explain how experimental data and your ratios in part (b) support the law of independent assortment.

7. In fruit flies, long wings (*L*) are dominant to short wings (*l*) and gray body colour (*G*) is dominant to black body colour (*g*). If flies that are heterozygous for both traits are crossed and 256 offspring are produced, how many of these offspring would you predict to have each phenotype below?

a) long, gray

b) long, black

c) short, gray

d) short, black

8. Imagine that you randomly select one of the long-winged gray-bodied flies from the offspring described in question 7 and perform a test cross with a fly that is homozygous recessive for both traits. What is the probability that the offspring will be produced in the ratio 1:1:1:1 for long, gray : long, black : short, gray : short, black?

9. Now imagine that you performed the cross described in question 8. Your results were 139 flies with long wings and gray bodies, 49 flies with long wings and black bodies, 53 flies with short wings and gray bodies, and 15 flies with short wings and black bodies. Are these the results you would expect? Explain your reasoning.

10. Explain how incompletely dominant and co-dominant alleles differ from dominant and recessive alleles. What type of observation would indicate that alleles are co-dominant?

11. A farmer crosses a black rooster with a white hen. Of the seven offspring, three are black, three are speckled black and white, and one is white.

a) What could you infer about the black and white alleles?

b) Do the ratios of characteristics among the offspring follow the pattern you would expect? Why or why not? What further information would you need in order to draw conclusions about the inheritance of this trait?

12. Describe the observations that suggested to scientists that genes might be carried on chromosomes.

SECTION 17.2

Extending Mendel's Laws: More Patterns and Probabilities

Section Outcomes

In this section, you will

- **explain** inheritance patterns for genes on the same chromosome
- **analyze** crossing over data and **create** a chromosome map for genes on a single chromosome
- **describe** inheritance patterns for sex-linked genes
- **compare** ratios and probabilities of genotypes and phenotypes for multiple alleles and for polygenic traits
- **design** and **perform** an experiment to investigate the influence of environmental variables on the expression of genetic information in an individual

Key Terms

linked genes
crossing over
chromosome mapping
map unit
recombinant types
parental type
recombination frequency
sex-linked traits
Barr body
multiple alleles
order of dominance
continuous traits
polygenic traits

A few years after Sutton suggested that genes are located on chromosomes, an American biologist named Thomas Hunt Morgan (see Figure 17.15) moved his research team into another laboratory at Columbia University. Morgan did not accept Sutton's arguments and intended to collect evidence for an alternative theory. For his research, he required an organism that was economical to maintain, could be raised in his small laboratory, and would reproduce rapidly. He chose the fruit fly, *Drosophila melanogaster*, and his laboratory became known as "the fly room." Morgan's research was characterized by rigorous experimental procedures and careful statistical analysis. His results soon convinced him of Sutton's chromosome theory and established much of the framework for today's genetic research.

In 1910, Morgan discovered an unusual white-eyed male among his fly population. He crossed the white-eyed male with a normal red-eyed female. All the F_1 generation had red eyes. This seemed to indicate that normal red eyes are dominant to the white-eye mutation. When Morgan crossed a male and female from the F_1 generation, however, the results surprised him. All the females of the F_2 generation had red eyes, while half the F_2 males had red eyes and half had white eyes. The discovery that eye colour was connected to gender led Morgan to deduce that the gene for eye colour is located on the X chromosome. This was the first gene to be mapped to a specific chromosome. Morgan went on to uncover additional experimental evidence for the chromosome theory.

Figure 17.15 Thomas Hunt Morgan (1866–1945) sought to disprove the chromosome theory of inheritance. Like all good scientists, he reconsidered his position when his experimental evidence did not support his initial hypothesis. In 1933, Morgan received the Nobel Prize for his work.

Linked Genes and Chromosome Maps

In Section 17.1, you learned that the movement of chromosomes follows the pattern that Mendel predicted for genes (what he called "factors"). This observation led to the chromosomal theory of inheritance: genes are carried on chromosomes, and chromosomes segregate independently into gametes during meiosis.

Sutton predicted that when alleles of two different genes are on the same chromosome they do not assort independently. For this reason, genes that are found on the same chromosome are sometimes called **linked genes**. Experimental data show, however, that linked genes segregate on a regular basis. How can alleles of two genes on the same chromosome be found in different gametes? The answer is found in the process of crossing over.

Crossing Over and Inheritance

In Chapter 16 you saw that crossing over occurs in prophase I of meiosis, when the non-sister chromatids in a tetrad exchange pieces of chromosomes. (Turn to page 564 and 566 for review.) Suppose that you are studying two genes. If the point at which a crossover occurs is between these genes, the alleles will be

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FYI

Morgan initially requested research funding to conduct his experiments on rabbits. His request for funding was denied, so Morgan used the cheaper—and faster-reproducing—*Drosophila* instead. Scientists now suggest that if Morgan had worked with rabbits, his breakthrough discoveries would have been delayed by about 25 years.

on separate chromosomes and will therefore migrate into different gametes.

Crossing over is a random event and occurs, with equal probability, at nearly any point on the sister chromatids, except near the centromere. This means that a crossover is more likely to occur between genes that are farther apart on a chromosome than between genes that are closer together.

In repeated experiments, Morgan and his students found that any given pair of linked genes would separate with a predictable frequency. They also found that this frequency varied among different pairs of linked genes. One of Morgan's students, Alfred Sturtevant, was the first to demonstrate that these results could be explained by assigning each gene a specific position along a linear chromosome. Because of Sturtevant's work, Morgan and his team amended the chromosome theory of inheritance. The *gene-chromosome theory* now states that *genes exist at specific sites arranged in a linear manner along chromosomes*.

Chromosome Mapping

The concept of crossing over is used to determine the relative positions of genes on a chromosome in a process called **chromosome mapping**. One **map unit** is defined as the distance between points on a chromosome where a crossover is likely to occur in 1 percent of all meiotic events. The *map distance* is the distance between genes on a single chromosome.

Imagine that you are a biologist working with Morgan in the famous "fly room" at Columbia University. As a researcher, you decide to perform a cross with *Drosophila melanogaster*. You cross a homozygous recessive purple-eyed, vestigial-winged fly (*ppvv*) with a heterozygous dominant normal-eyed, normal-winged fly (*PpVv*). You expect the phenotype ratio for the cross to be 1:1, because both genes are found on chromosome 2. To your surprise, you find that 45 percent of the flies have normal eyes and normal wings, 45 percent have purple eyes and vestigial wings, 5 percent have normal eyes and vestigial wings, and 5 percent have purple eyes and normal wings. After discussing your results with your colleagues, you realize that for a fly in the F_1 generation to have the phenotypes purple eyes and normal wings, or normal eyes and vestigial wings, a crossover must have occurred during meiosis. Since the chromosomes of these F_1 flies have a different combination of alleles than the chromosomes of the P generation, these flies are called **recombinant types**, or recombinants. The F_1 flies with the phenotypes normal eyes and normal wings or purple eyes and vestigial wings are called **parental types** because their chromosomes are identical to those of the P generation.

As you know, the greater the distance is between linked genes, the more likely they are to cross over during meiosis. In fact, the percentage of recombinant types in the F_1 generation is directly proportional to the distance between the genes. The percentage of flies that are recombinant types corresponds to the **recombination frequency**—that is, the percentage of times that a crossover occurred as P gametes were formed.

Now imagine that your cross produces 1000 offspring. Of these offspring, 450 of have normal eyes and normal wings, 450 have purple eyes and vestigial wings, 50 have normal eyes and vestigial wings, and 50 have purple eyes and normal wings. The recombination frequency would be determined as follows:

$$\text{Recombination frequency} = \frac{50 + 50}{1000} \times 100\% = 10\%$$

BiologyFile

FYI

The map unit has been given the name *centimorgan* to honour Morgan's contribution to the chromosomal theory of inheritance.

$$\text{Recombination frequency} = \frac{\text{number of recombinant types}}{\text{total number of offspring}} \times 100\%$$

Thus, in your cross, 10% of the F_1 generation were recombinant types. Because the recombination frequency is directly proportional to map distance (1% = 1 map unit), you can use this value to create a chromosome map showing the relative distances between the linked genes. For example, in your cross, the genes for eye colour and wing type are 10 map units apart.

After conducting further crosses, you discover that the map distances between three different genes on chromosome 2 are as follows:

- The distance between the genes for eye colour and wing type is 10 map units.
- The distance between the genes for eye colour and body colour is 4 map units.
- The distance between the genes for body colour and wing type is 6 map units.

How does this information help you determine the order in which the genes are found on chromosome 2? In order to fit the data, the gene for eye colour must be 4 map units from the gene for body colour and 10 map units away from the gene for wing type. Thus, in order for the gene for body colour to be 6 map units away from the gene for wing type, it must fall between the other two genes as shown in Figure 17.16.

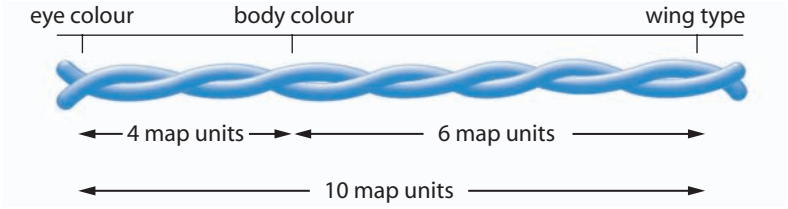


Figure 17.16 Mapping chromosomes. Genes are arranged linearly on the chromosome at specific gene loci.

Q 18 Differentiate between the chromosome theory of inheritance and the gene-chromosome theory.

Q 19 What is chromosome mapping?

Q 20 What is a map unit?

Q 21 Distinguish between the terms recombinant types and parental types.

Sex-Linked Inheritance

As you know, the first gene to be mapped to a specific chromosome was the gene coding for white eye colour in *Drosophila melanogaster*. Once Morgan realized that this gene is located on the X chromosome, he found that the unusual inheritance pattern could be explained by simple Mendelian genetics.

Like humans, female fruit flies have two X chromosomes, while males have one X chromosome and one Y chromosome. The F_1 data indicated that the white-eye trait is recessive. Therefore, to have white eyes, a female fruit fly must inherit an X chromosome with the recessive allele from each parent. A male fruit fly, however, has to inherit only a single recessive allele from the female parent to display the white-eye trait because it has only one X chromosome. (The male parent donates the Y chromosome, which does not carry the allele.)

Traits that are controlled by genes on either the X or Y chromosome are called **sex-linked traits**. Punnett squares can be used to predict the outcome of crosses that involve sex-linked traits. Figure 17.17 shows a cross between a wild-type female and a white-eyed male.

In humans, a common form of colour blindness is the result of a recessive trait that is carried on the X chromosome. This trait makes it difficult to distinguish between reds and greens.

BiologyFile

Try This
Use a Punnett square to predict the genotype and phenotype ratios of the F_2 generation of fruit flies, crossing a male and a female from the F_1 generation.

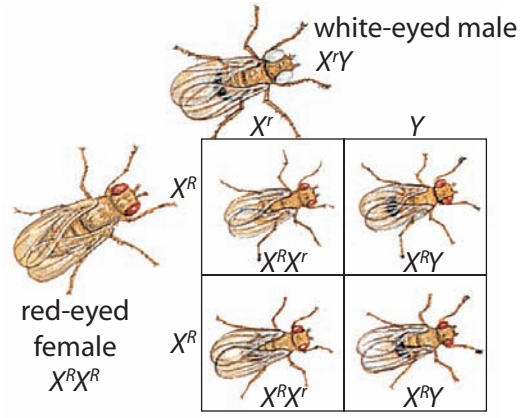


Figure 17.17 A cross of fruit flies, showing the inheritance of alleles on the X chromosome. What is the expected ratio of male to female offspring from this cross?

Thought Lab 17.1 Mapping Chromosomes

Target Skills

Analyzing crossover data to create a chromosome map

In Part A of this activity, you will follow a procedure to construct a chromosome map. In preparation for future studies of genetics, your teacher may choose to assign Part B to expose you to the special notation geneticists use for analyzing linked genes.

Part A: Constructing a Chromosome Map

Procedure

1. Using the table below as a guide, determine the map distance between the linked genes for eye colour and wing type in the following experiment.

   You perform the following cross: *PpVv* × *ppvv*. You count 1000 offspring in the F_1 generation. You find that 406 of the flies have normal eyes and normal wings, 398 have purple eyes and vestigial wings, 96 have normal eyes and vestigial wings, and 100 have purple eyes and normal wings.

2. From previous research findings, you know that the distance between the gene for eye colour and the gene for body colour is 12.2 map units, and the gene for body colour is 7.4 map units away from the gene for wing type. All three genes are on the same chromosome. Draw a chromosome map showing the relative distances between the three linked genes.

Analysis

1. You conduct the same cross again, but this time you get an almost exact 1:1 ratio of flies with normal eyes and normal wings to flies with purple eyes and vestigial wings. There are no recombinant types. Provide two explanations that might account for these results.

2. Linkage data has been used to map genes on the chromosomes of *Drosophila melanogaster*. Do you think such data could be used to map human chromosomes? Explain your reasoning.

Part B: Using Linked Gene Notation

In order to describe linked genes, fruit fly geneticists developed a different notation for genes. The normal allele—the allele that is nearly always found in natural populations—is called the *wild type* allele. The wild type allele is represented with a plus sign (+). The mutant allele for a gene is represented with lower-case italic letters. Linked genes are represented with a horizontal line or a slash.

For example, the gene for purple eyes (*pr*) and the gene for vestigial wings (*vg*) are on chromosome number 2. The homozygous recessive genotype is represented as $\frac{prvg}{prvg}$ or *pr vg* / *pr vg* to indicate that these genes are on the same chromosome. The homozygous dominant genotype is represented as $\frac{+\ +}{+\ +}$ or + + / + +.

Procedure

1. Create a chromosome map of three linked genes based on the research presented below.

   a) In fruit flies, the mutant gene *d* causes short legs and the mutant gene *pr* causes purple eyes. A geneticist performs the following cross: *pr d* / + + × *pr d* / *pr d*. She counts 1000 offspring and finds 391 wild type, 115 purple-eyed and normal-legged, 105 normal-eyed and short-legged, and 389 purple-eyed and short-legged. What is the map distance between the genes for leg length and eye colour?

   b) The same geneticist then performs the following cross: *vg d* / + + × *vg d* / *vg d*. She counts 1000 offspring and finds 350 wild type, 154 vestigial-winged and normal-legged, 153 normal-winged and short-legged, and 343 vestigial-winged and short-legged. What is the map distance between the genes for leg length and wing type?

   c) The recombination frequency for the gene for eye colour and the gene for wing type is 8.7%. What is the map distance between these two genes? Draw a chromosome map showing the relative distances between all three linked genes studied in Part B.

Analysis

1. Do you think the linked gene notation used by geneticists simplifies or complicates linked gene analysis? Explain your reasoning.

Steps to determine map distance

Step	Example
Perform a cross between a fly that is known to be heterozygous for both traits and a fly that is homozygous recessive for both traits.	*PpVv* × *ppvv*
Collect a large number of F_1 flies from the cross. Determine the number of flies with each of the possible phenotypes.	Possible non-recombinant phenotypes: • purple eye, vestigial wing • normal eye, normal wing Possible recombinant phenotypes: • purple eye, normal wing • normal eye, vestigial wing
Calculate the total number of recombinant F_1 phenotypes as a percentage of the total number of F_1 flies. This gives you the recombination frequency and the number of map units separating the two genes.	$\frac{\text{number of recombinant types}}{\text{total number of offspring}} \times 100\%$

For example, people who are red-green colour blind cannot see the number in Figure 17.18. About 8 percent of men and 0.4 percent of women have this form of colour blindness. Figure 17.19 shows how this trait is passed on in families.

In humans and most other animals, only the male carries a Y chromosome. In these species, Y-linked traits are passed only from males to their sons. A female cannot be a carrier of a Y-linked trait. There are relatively few Y-linked traits. The gene for hairy ears is found on the Y chromosome.

Barr Bodies

An extra chromosome or a missing chromosome can result in a serious and even lethal genetic disorder. How, then, is it possible for females to carry two X chromosomes and males only one,

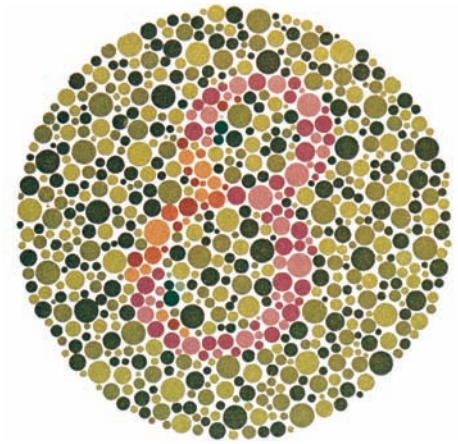


Figure 17.18 Can you see the number hidden in the dots? What can you conclude about your own genotype?

without any apparent difference in the expression of X-linked genes? The answer is that every cell has only one functioning X chromosome. In every female cell, one of the X chromosomes is inactive. The inactive X chromosome is condensed tightly into a structure known as a **Barr body**. Either of the two

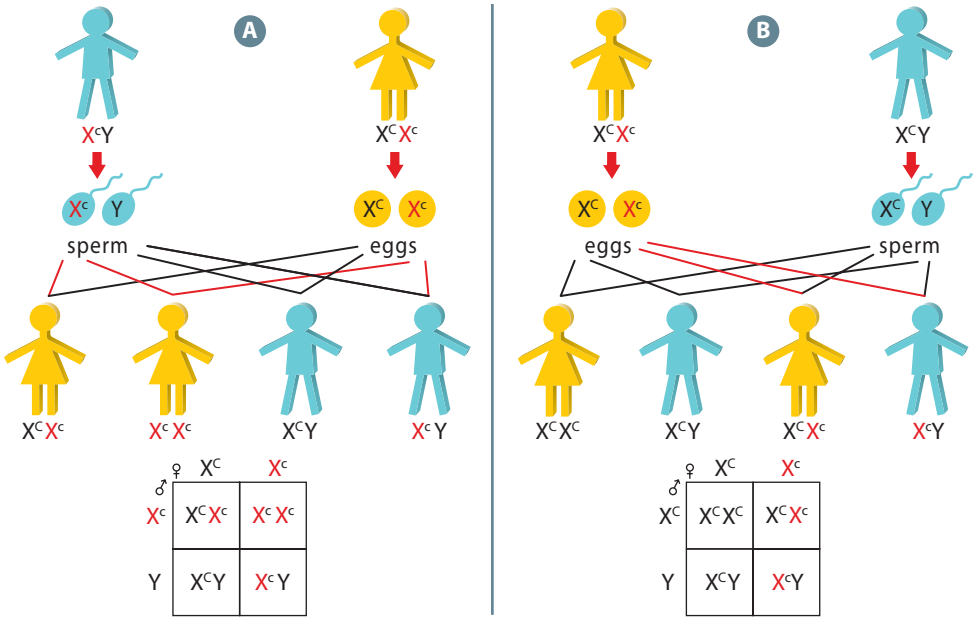


Figure 17.19 An allele for sex-linked colour blindness is passed to the next generation. Males can pass the X-linked recessive trait only to their daughters **(A)**. Females who are heterozygous for the condition have a 50 percent chance of passing the recessive allele to a child **(B)**. Can a man with normal vision have a child who is colour blind?

Practice Problems

8. A woman who has normal vision and the heterozygous genotype X^CX^c marries a man who is colour-blind (X^cY). What is the expected ratio of genotypes and phenotypes among their children?

9. Suppose that you have one wild-type female fly and one white-eyed male fly. What steps would you follow to produce a white-eyed female fly? Illustrate your steps with Punnett squares.

10. In a species of dog, a mutant gene that causes deafness is found on the Y chromosome. Draw a Punnett square to show the outcomes of a cross between

a) a male dog whose father is deaf and a female dog whose father is not deaf

b) a female dog whose father is deaf and a male dog whose father is not deaf

Figure 17.20 Male tortoiseshell cats are very rare. Why is this not surprising?

X chromosomes can be inactive—and which X chromosome forms a Barr body is random.

A visible effect of the inactivation of one X chromosome is the calico, or tortoiseshell, coat colour in cats, such as the type shown in Figure 17.20. The tortoiseshell coat colour is the result of a random distribution of orange and black patches. The gene that codes for coat colour (orange or black) is located on the X chromosome. (Some tortoiseshell cats also have white patches, but a different gene codes for these.) A tortoiseshell cat is heterozygous for the coat colour allele. That is, one X chromosome carries the allele for black fur, and the other X chromosome carries the allele for orange fur. At an early stage of the cat's embryonic development, one X chromosome in each cell is deactivated. The descendants of these cells have the same inactive X as their parent cells. When the kitten is born, patches of orange show collections of cells in which the X chromosome that is carrying the black allele is deactivated, and patches of black show collections of cells in which the X chromosome that is carrying the orange allele is deactivated.

22 What are sex-linked traits?

23 Explain why the formation of a Barr body means that human females have only one functioning X chromosome in each somatic cell.

24 Explain how the formation of a Barr body accounts for the coat colour of a tortoiseshell cat.

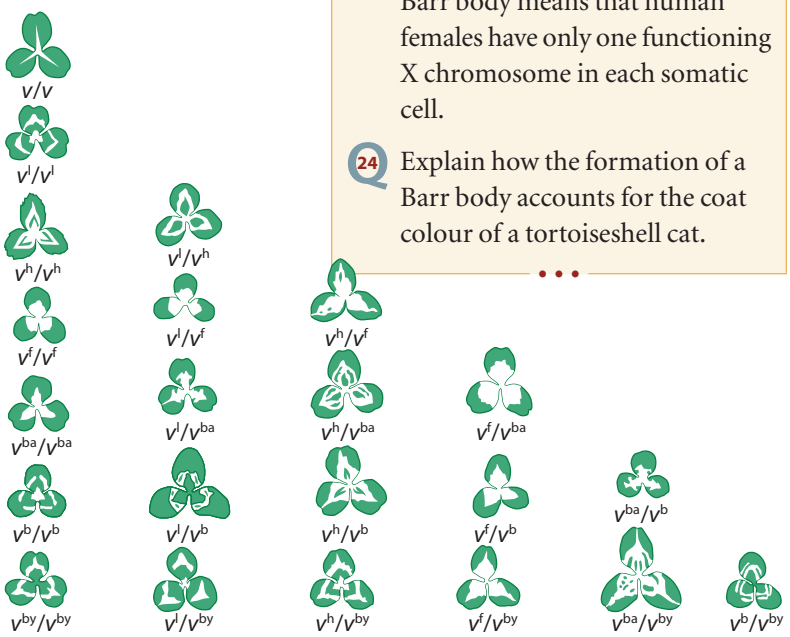


Figure 17.21 A single gene is responsible for each of the 22 patterns that can be expressed in clover leaves.

Multiple Alleles

You know that a maximum of three genotypes can be produced by one gene with two alleles. Two of the genotypes are homozygous for one of the two alleles, and the third genotype is heterozygous. Only one gene, however, is responsible for all the patterns on the leaves of the white clover (*Trifolium repens*) shown in Figure 17.21. How is this possible?

In clover, as in most other organisms, many genes have more than two alleles. A gene with more than two alleles is said to have **multiple alleles**. Any individual organism has only two alleles for each gene (one allele on each homologous chromosome), but many different alleles exist within the population as a whole. If you examine the genotypes below each phenotype in Figure 17.21, you can see that there are seven different alleles for cloverleaf patterns.

In humans, a single gene determines a person's ABO blood type. This gene determines the type of antigen, if any, that is attached to the cell membrane of red blood cells. The gene is designated *I*, and it has three common alleles: I^A, I^B, and *i*. The different combinations of the three alleles produce four different phenotypes, which are commonly called blood types: A, B, AB, or O. (Refer to Figure 17.22.) The I^A allele is responsible for the presence of

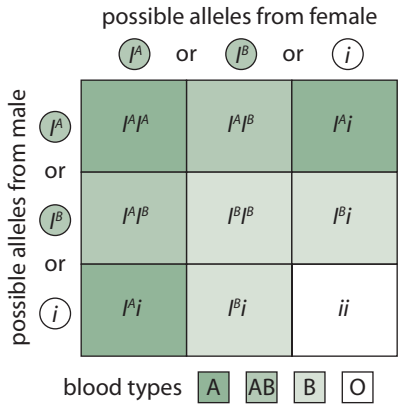


Figure 17.22 Different combinations of the three *I* gene alleles result in four different blood type phenotypes: type A (either I^AI^A homozygotes or I^Ai heterozygotes), type B (either I^BI^B homozygotes or I^Bi heterozygotes), type AB (I^AI^B heterozygotes) and type O (*ii* homozygotes).

Table 17.2 ABO Blood Types

Genotype	Phenotype	Antigen
ii	O	none
I^Ai	A	A
I^AI^A	A	A
I^Bi	B	B
I^BI^B	B	B
I^AI^B	AB	A and B

an A antigen on the red blood cells. The I^B allele is responsible for the presence of the B antigen, and the i allele causes there to be no antigen. Of the three possible alleles that determine blood type, one (i) is recessive to the other two, and the other two (I^A and I^B) are co-dominant with each other. Table 17.2 shows blood genotypes, along with their phenotypes and the antigens on the red blood cells.

Another well-researched example of multiple alleles involves coat colour in rabbits, as shown in Figure 17.23. The gene that controls coat colour in rabbits has four alleles: agouti (C), chinchilla (c^{ch}), Himalayan (c^h), and albino (c). In the order given, each allele is dominant to all the alleles that follow. The **order of dominance** sequence can be written as $C > c^{ch} > c^h > c$, where the symbol > means "is dominant to."

25 What are multiple alleles?

The Sample Problem on page 606 will help you learn how to analyze crosses that involve multiple alleles. Study the problem-solving strategy carefully, and then use it to solve the Practice Problems that follow.

Polygenic Inheritance

Plant height was one of the traits that Mendel studied. He carefully selected plants that had strikingly different heights so there would be no question about phenotypes when he counted offspring. After Mendel worked out the basic mechanisms of inheritance, other people began to look at **continuous traits**—traits for which the phenotypes vary gradually from one extreme to another. Some examples of continuous traits include height in humans, ear length in corn, kernel colour in wheat, and weight of beans. Continuous traits cannot be placed into discrete categories (such as "colour blind" and "not colour blind"). Instead, they vary over a continuum. For example, humans cannot simply be categorized as "short" and "tall." Instead, height in humans

BiologyFile

Try This
Recall your studies of evolution and speciation in Chapter 4. How could polygenic inheritance contribute to the processes of evolution and speciation?

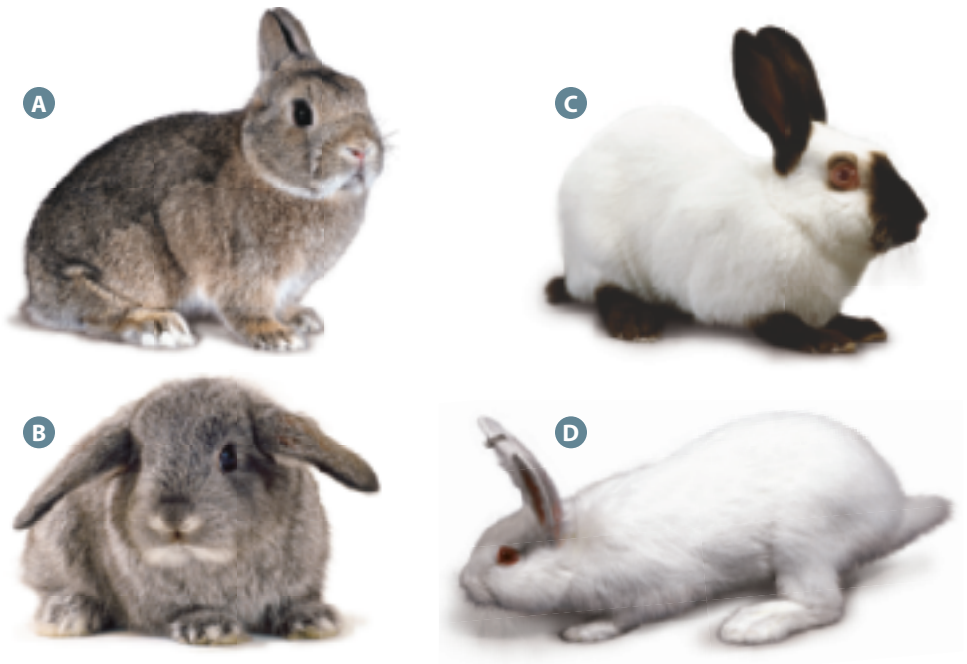


Figure 17.23 The agouti rabbit **(A)** could have genotypes CC, Cc^{ch}, Cc^h, or Cc. The chinchilla rabbit **(B)** could have genotypes $c^{ch}c^{ch}$, $c^{ch}c^h$, or $c^{ch}c$. The Himalayan rabbit **(C)** could have genotypes c^hc^h or c^hc. The albino rabbit **(D)** must have the genotype cc.

Sample Problem

Human Blood Types

Problem

If a man has type O blood and a woman has type B blood, what possible blood types could their children have? If this couple has six children, all with type B blood, what could you state about the woman's genotype?

What Is Required?

Possible blood types of children
Statement about woman's genotype

What Is Given?

The man has type O blood.
The woman has type B blood.

Plan Your Strategy

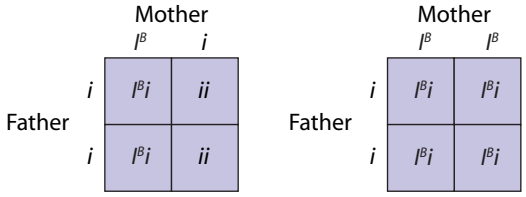
Step 1 Determine the possible genotypes of the man and the woman.
Step 2 Make Punnett squares for all the possible combinations of genotypes.
Step 3 List all the possible genotypes of the children.
Step 4 State all the possible phenotypes (blood types) produced by these genotypes.

Act on Your Strategy

Step 1

The man must have the genotype *ii*.
The woman could have genotype I^Bi or I^BI^B.

Step 2



Step 3

Children could have genotypes I^Bi or *ii*. These genotypes produce type B and type O blood, respectively.

Step 4

If the couple has six children with type B blood, you would suspect that the woman's genotype is I^BI^B. You could not be certain, however, because the processes involved in gamete formation are random. The woman could have the genotype I^Bi, but none of the children received the *i* allele. (If the woman has the genotype I^Bi, the chances of all six children receiving her I^B allele are 1 in 64, or about 1.6 percent.)

Check Your Solution

The only genotype that produces type O blood is *ii*. To produce type B blood, the woman must have at least one I^B allele. Her second allele could be *i* or I^B. All the children had to receive an *i* allele from their father. They could receive either an *i* or an I^B allele from their mother.

Practice Problems

11. If a man has type AB blood and a woman has type A blood, what possible blood types could their children have?

12. In one family, all three siblings have type B blood.

a) Use Punnett squares to show how two different sets of parent genotypes could produce this result.

b) Which of the two sets of potential parents in your answer to (a) is more likely to be the parents of these siblings?

13. A couple just brought home a new baby from the hospital. They begin to believe that the hospital switched babies, and the baby they brought home is not theirs. They check the hospital records and find that the man's blood type is B, the woman's blood type is AB, and the baby's blood type is O. Could the baby be theirs?

14. A chinchilla rabbit with genotype $c^{ch}c^h$ is crossed with a Himalayan rabbit with genotype c^hc. What is the expected ratio of phenotypes among the offspring of this cross?

15. Some of the offspring of a chinchilla rabbit and a Himalayan rabbit are albino. What must be the genotypes of the parent rabbits?

16. Could a mating between a chinchilla rabbit and an albino rabbit produce a Himalayan rabbit? Explain your reasoning, with reference to the genotypes and phenotypes of the parents and possible offspring.

17. Four children have the following blood types: A, B, AB, and O. Could these children have the same two biological parents? Explain.

varies over a wide range of values. Thus, it is a continuous trait. How is it possible for discrete elements, such as genes, to control a gradually changing trait?

Continuous traits are usually controlled by more than one gene. It is often difficult to determine the precise number of genes that control a given trait, but there could be as many as five or more for some traits. Traits that are controlled by many genes are called **polygenic traits**. A group of genes that all contribute to the same trait is called a *polygene*. Each dominant allele contributes to the trait—for example, adds to a person's height. Recessive alleles do not contribute to the trait.

To see how polygenes generate offspring with a continuous trait, consider ear length in corn. Assume that ear length is controlled by two genes, *A* and *B*. In true-breeding corn with the genotype *AABB*, four dominant alleles contribute to ear length. As a result, this genotype has the longest ears. True-breeding corn with the genotype *aabb* has four recessive alleles, none of which contribute to ear length. Thus, this genotype has the shortest ears. If true-breeding lines for shortest ears of corn and longest ears of corn are crossed, the F_1 generation will have medium-length ears (*AaBb*) where two dominant alleles contribute to ear length.

P $AABB \times aabb$

F_1 $AaBb$

The Punnett square for the F_2 generation is shown in Figure 17.24. With just two genes, creating a range of zero to four contributing alleles, you start to see a distribution of ear lengths. If there are three genes (*A*, *B*, and *C*) creating a range of zero to six contributing alleles, the ratio of phenotypes is 1:6:15:20:15:6:1, as shown in bar graph C in Figure 17.24. As you can see, when more genes contribute to ear length, the curve representing ear length becomes more gradual. Environmental conditions, such as temperature, precipitation, and soil composition, also influence ear length in corn. The overall result is a very gradual curve representing ear length.

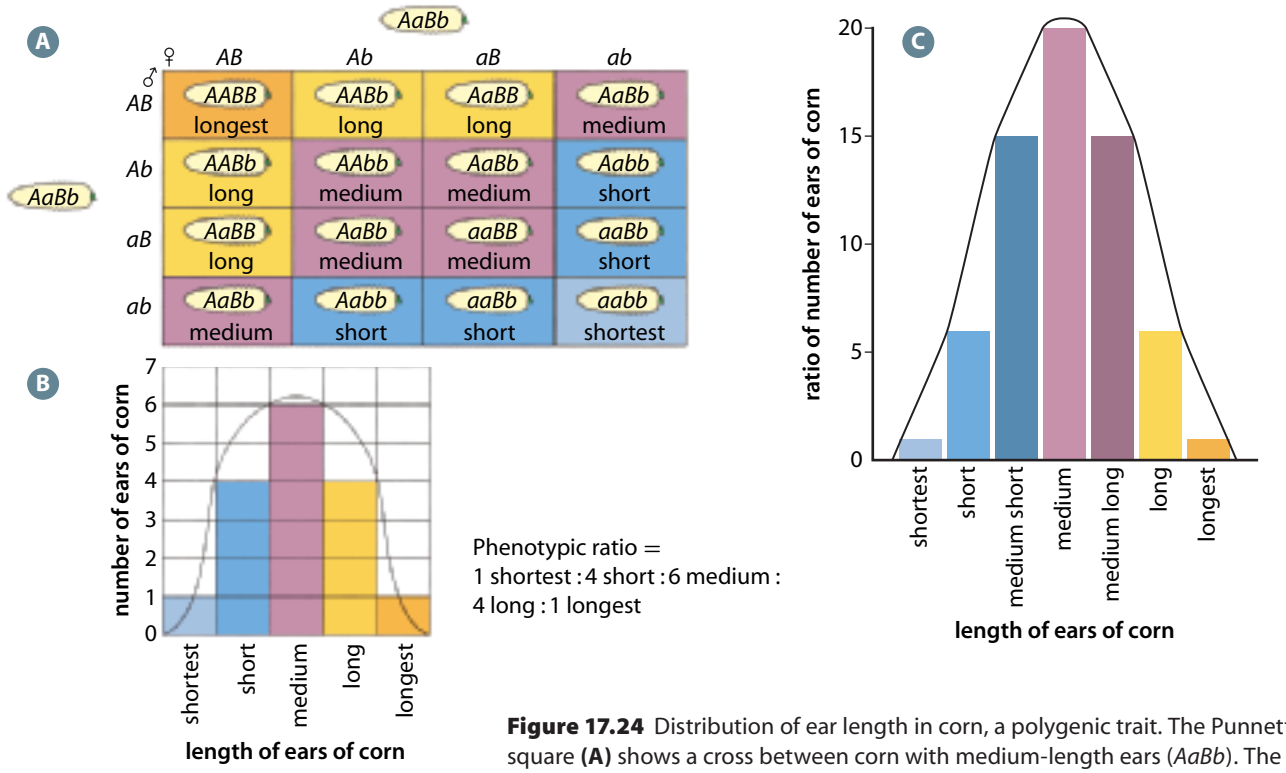


Figure 17.24 Distribution of ear length in corn, a polygenic trait. The Punnett square **(A)** shows a cross between corn with medium-length ears (*AaBb*). The resulting phenotype ratio of 1:4:6:4:1 is graphed in **(B)**. In graph **(C)**, three genes control ear length, resulting in a more gradual-length distribution curve.

INVESTIGATION 17.B

Target Skills

Designing and **performing** an experiment to demonstrate a causal relationship between an environmental factor and the expression of genetic information in plants

Environmental Influences on Gene Expression

As you know, chlorophyll is the molecule that allows plants to capture energy from sunlight and use this energy to produce nutrients. Chlorophyll is also the pigment that gives leaves their green colour. Plants that produce chlorophyll appear green, while plants that do not produce chlorophyll appear pale yellow.

You will work in small groups for this investigation. Using the materials provided, your group will design and conduct an experiment to test the influence of light on the production of chlorophyll. Your experiment must enable you to draw conclusions about

- the minimum duration of exposure to light required to trigger the production of chlorophyll
- whether the triggering event is reversible (that is, whether chlorophyll production starts and stops as environmental conditions change)

Question

How does light influence the production of chlorophyll in germinating plants?

Hypothesis

Formulate a hypothesis to explain how light influences the expression of the genes responsible for chlorophyll production. You will use this hypothesis as the basis of your experimental design.

Safety Precautions

Wash your hands after handling materials in the laboratory.

Materials

- seeds (All seeds carry the same genes for chlorophyll production.)
- petri dishes
- labels
- light source
- shoe boxes
- water
- paper towels
- graduated cylinder

Experimental Plan

1. Brainstorm several methods you could use to test your hypothesis. As a group, select one method for your experimental design.
2. Your experimental design should include the collection of qualitative and quantitative data.
3. Identify the responding and manipulated variables. Which variables do you need to control?
4. As you prepare your procedure, be sure to consider the time required for each step.
5. Prepare the data table you will use to record your observations. Decide what form (such as a chart or graph) you will use to present your results.
6. Review your procedure with your teacher. When your procedure has been approved by your teacher and all the members of your group, you are ready to begin.

Data and Observations

7. Record your observations in your table. Make notes about any findings that do not fit in your data table. Record any questions that come up as you conduct your experiment.

Analysis

1. Did your observations support your hypothesis? Explain.
2. Did your experiment allow you to draw conclusions about the inheritance of chlorophyll-producing genes? Why or why not?
3. Identify the variables you considered when designing your experiment. Explain why you needed to consider each variable to obtain scientifically valid results.

Conclusions

4. State your conclusions about the relationship between exposure to light and expression of chlorophyll-producing genes.
5. Could a different hypothesis also explain the results of your experiment? How could you design an experiment to test this different hypothesis?

Extensions

6. What social benefit could come from understanding the effect of light on chlorophyll production?

Q26 What is the difference between a trait that has multiple alleles and a trait that is controlled by multiple genes? Give an example of each.

Q27 What is a continuous trait?

Genes and the Environment

Environmental conditions often affect the expression of genetic traits. For example, some genes are influenced by temperature. The colour pattern of Himalayan rabbits (Figure 17.23) is similar to the colour pattern of Siamese cats (see Figure 17.25). Their fur is pigmented on the cooler parts of their bodies: the face, ears, tails, and feet. In these animals, dark colouring is the result of a gene that is only active below a certain temperature. Another effect of a gene influenced by temperature can be seen in fruit flies. The expression of a mutant form called curly wings depends on temperature. If flies that are homozygous for curly wings are raised at 25 °C, their wings will be curly. If they are raised at 16 °C, their wings will be straight.

Q28 Give an example of one way (other than the ways described above) that environment can influence the expression of genetic traits.

Section 17.2 Summary

- Several variables that extend classical Mendelian genetics.
- These variables produce patterns of inheritance and phenotypes that do not, at first, appear to be consistent with Mendel's laws. Each is consistent, however, with the gene-chromosome theory.
- Sex-linked traits are expressed in different ratios by male and female offspring, because they are governed by the segregation of X and Y chromosomes.
- Linked genes do not segregate independently. Instead, the probability of recombination is determined by the relative positions of the alleles on the chromosome.
- The frequency of recombination can be used to construct a chromosome map.
- Other inheritance patterns that extend Mendel's laws are those for traits that are influenced by more than one gene or by environmental conditions, as well as those with multiple alleles.

Figure 17.25 Siamese and Burmese cats have an allele that produces a dark fur colour. All the cells have the same allele, but it is expressed only in cells whose temperature is lower than about 33 °C.

Section 17.2 Review

1. Describe how the process of crossing over of non-sister chromatids led to an understanding of linked genes.

2. A woman with normal vision marries a man with normal vision. They have three children, and one is colour blind.

a) What can you conclude about the genotypes of the parents?

b) What sex is the child who is colour blind? How do you know?

3. Could a person with type AB blood have a child with type O blood? Explain.

4. Your friend keeps rabbits as pets. She has bred her female albino rabbit with her male Himalayan rabbit. "I'm hoping I'll get some agouti rabbits," she says. What are her chances of getting an agouti rabbit? Explain.

5. In one species of bean plant, weight is influenced by two different genes.

a) How many weight classes would you expect to find in this plant population? Explain using a Punnett square or another visual representation.

b) When you analyze the weights of several beans from the same cross, you find that many beans have weights between the predicted weight classes. Identify two other factors that could influence the bean phenotypes.

6. Siamese cats that spend their lives indoors tend to have lighter-coloured fur than Siamese cats that live outdoors. What genetic process could account for this change?

SECTION 17.3

Genetics and Society

Section Outcomes

In this section, you will

- **describe** ways in which plant and animal breeding programs make use of genetic research
- **draw** and **interpret** pedigree charts that show the inheritance of single-allele, sex-linked and multiple-allele traits in humans
- **design** and **collaborate** on a plan to investigate the inheritance of human traits
- **assess** the role of genetic counselling and technology in issues that involve society
- **evaluate** some of the social, ethical, and economic considerations that are involved in the application of genetic research

Key Terms

pedigree
genetic screening
genetic counsellor

Figure 17.26 Farmers in all three Prairie provinces grow large quantities of canola (*Brassica napus*). In Alberta, close to one million hectares of cropland are seeded with canola each year.

The field of genetics, as it is known today, is a relatively recent scientific discipline, originating with Gregor Mendel. However, genetics is rooted—literally—much farther in the past, in the earliest practices of agriculture. Traditional agriculture involves the controlled breeding of plants and animals with specific combinations of useful or desirable inherited phenotypes. Traditional agriculture is often imprecise, because it combines many genes (and, therefore, many traits) at a time. Nevertheless, in the hands of skillful, observant, and patient farmers and breeders, selective breeding has resulted in the Appaloosa (recall the chapter introduction) and many of the plant and animal products on which modern society depends.

Breeding Plants

About 7000 years ago, in the area that is now southern Mexico, Native American peoples were breeding maize (more commonly called corn, *Zea mays*). The evolution of maize, which became a staple in their diet, required thousands of generations of selecting and breeding the plants that produced the largest grains and the most clusters of grain. For years, biologists thought that maize was descended from a plant called teosinte (*Zea mexicana*). Studies involving both genetics and archeology now cast doubt on this hypothesis. Maize is related to teosinte, but may have resulted from a cross involving another source of genes from gamagrass (*Tripsacum* sp.). In either case, modern maize is the product of thousands of years of careful breeding.

The development of canola (see Figure 17.26) by plant breeders at Agriculture Canada's Research Centre in Saskatoon is a more recent example of a success story involving selective breeding. Oil from the oilseed plant, also called rapeseed, was long used as a lubricant. Since two chemicals that are found in rapeseed oil—erucic acid and glucosinolates—are toxic to laboratory animals, rapeseed oil was banned for human consumption. In the 1970s, plant scientists in Saskatoon carried out a breeding program that successfully developed a variety of rapeseed with such low levels of erucic acid and glucosinolates that the oil has been approved for human consumption.

The new variety was named *canola*, in recognition of its Canadian origin. Canola is considered to be one of the healthiest vegetable oils available because it is high in monounsaturated and polyunsaturated fats—factors that decrease the likelihood of plaque-forming deposits in arteries. Canola is now grown and used worldwide.

Breeding Animals

Historical records show that the Shorthorn cow, shown in Figure 17.27, first became a recognized breed about 400 years ago in Northern England. This makes the Shorthorn one of the world's oldest established breeds of domestic cattle. Shorthorn cows were first imported to North America in the mid-1800s, and they quickly became popular as both dairy and beef cattle.

Research teams continue to study ways to improve the Shorthorn's traits. Some estimates suggest that the average milk production of Canadian Shorthorn herds increased by almost 30 percent between 1990 and 2000. Successful breeding programs such as this rely on an understanding of Mendelian genetics. For example, although bulls do not produce milk, they can carry genes that make their female offspring good milk producers. Animal breeders can use the characteristics of offspring to deduce which bulls carry these genes.

Similar breeding programs have been used in the Canadian beef industry. At one time, a steak was considered to be good if it had a lot of "marbling" of fat. With current knowledge that animal fat is not as healthy for the diet as lean meat, cattle have been bred to be leaner. Pigs have also been bred to produce leaner meat. When nutritionists began to emphasize the dangers of consuming too much cholesterol, chicken breeders developed chickens that produce eggs with less cholesterol. Thus, knowledge of both nutrition and animal genetics has made it possible to breed animals that provide larger amounts of healthier products.

Figure 17.27 The Canadian Milking Shorthorn is found on many dairy farms on the Prairies. What steps would you take to find out if this bull's female offspring are likely to be good milk producers?

Q29 Give an example (other than those provided in the textbook) of how selective breeding has developed a new species of plant or animal.

Human Genetics

In the previous two sections, you have learned that the controlled breeding of plants and animals was the basis for many breakthroughs in genetics research. This kind of approach, however, does not work in the study of human genetics. It is clearly impossible for researchers to perform experimental crosses between selected men and women. As well, researchers cannot accumulate large numbers of offspring from the same parents in order to improve the statistical reliability of their data. Instead, when geneticists want to learn about the inheritance of human traits, they collect as much information about a family's history as they can and use this information to create a diagram called a pedigree. A **pedigree** is a type of flowchart that uses symbols to show the patterns of relationships and traits in a family over many generations (see Figure 17.28 on the next page).

Analyzing a Human Pedigree

You can use a pedigree to determine the pattern of inheritance of a particular trait. Examine the pedigree in Figure 17.29 on the next page.

BiologyFile

Web Link

The Newfoundland Pony is a breed of horse that arose during the history of settlement in Newfoundland. What horses are part of the genetic heritage of this breed? What environmental factors influenced the development of this breed? What factors led to its decline, and what is its current population?

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WWW

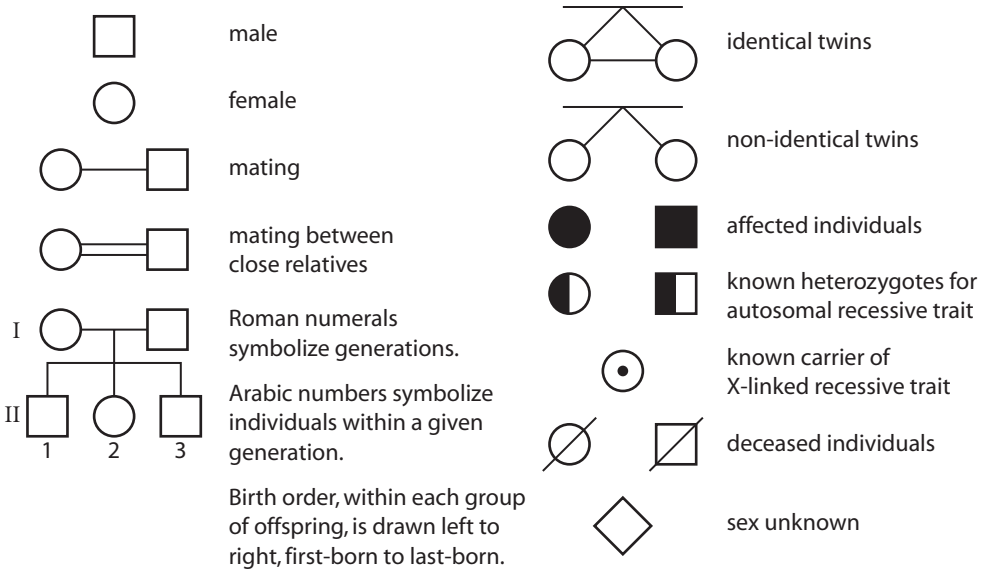


Figure 17.28 To make comparisons between different pedigrees as easy as possible, geneticists use several symbols to prepare a pedigree.

Autosomal Dominant Inheritance

Autosomes are any chromosomes other than sex chromosomes. *Autosomal inheritance* refers to traits—dominant and recessive—that are coded for by genes on autosomes. Figure 17.29 shows a human pedigree for an autosomal dominant condition called polydactyly—the occurrence of extra fingers or toes. Notice that both parents in the first generation are polydactylous. Two of their three children are also polydactylous. One child is not. Whenever a recessive phenotype occurs in a child of parents who exhibit the dominant trait, the parents must be heterozygous for that trait. Therefore, the normal allele must be recessive, which means that polydactyly is the dominant allele.

Huntington's disease is another autosomal dominant condition. It is a lethal disorder in which the brain deteriorates over a period of about 15 years. Its symptoms usually first appear after age 35, which is often after the affected individual has already had children. Early symptoms include irritability and memory loss, along with involuntary muscle movements. Over time, these symptoms become more severe, resulting in dementia and loss of muscle control.

Marfan syndrome is an autosomal dominant condition that affects the connective tissues. Because these tissues are found throughout the body, the disorder can affect all of the body's systems. Some common symptoms are unusually long bones, which can lead to abnormal curvature of the spine; eye problems, such as glaucoma, and an increased risk of retinal detachment; faulty heart valves; and respiratory problems. Symptoms tend to be mild in young people, but they become increasingly severe over time.

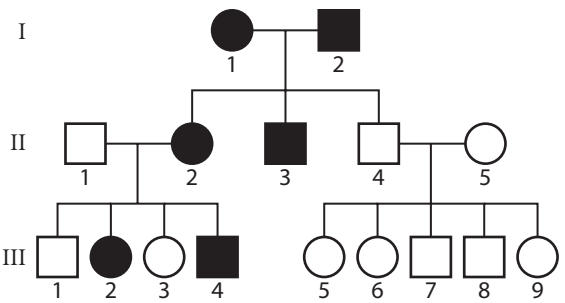


Figure 17.29 This pedigree shows the inheritance of polydactyly—an autosomal dominant trait. Notice that heterozygotes are affected by the trait, and that an affected child must have at least one affected parent. How do you know that individual II 2 is heterozygous?

Q30 Distinguish between the meaning of roman numerals and arabic numerals in a pedigree.

Q31 What is autosomal inheritance?

Autosomal Recessive Inheritance

Figure 17.30 shows a pedigree for an autosomal recessive disorder called phenylketonuria (PKU), which affects the development of the nervous system.

In people with PKU, an enzyme that converts phenylalanine to tyrosine is defective or absent, causing phenylalanine to convert to phenylpyruvic acid, which builds up to toxic levels. (Tyrosine is used by the body to make melanin and certain hormones.) Babies with PKU appear healthy at birth. If their condition is not diagnosed and treated, however, they will become severely mentally handicapped within a few months. Newborns are routinely tested for PKU. If they test positive for the disorder, they are placed on a very restrictive, phenylalanine-free diet. The dietary restrictions can be eased later in life, once the nervous system is more fully developed.

Another autosomal recessive disorder is cystic fibrosis. Cystic fibrosis causes a buildup of thick mucus in the lungs and digestive system. People with cystic fibrosis have an increased risk of pneumonia and respiratory failure, as well as difficulty digesting their food. The treatment includes a combination of physical therapy, nutritional supplements, and antibiotics. An estimated one in 2500 Canadian children is born with cystic fibrosis, making this the most common lethal genetic condition affecting young Canadians. Scientists estimate that one in 25 Canadians carries the recessive gene.

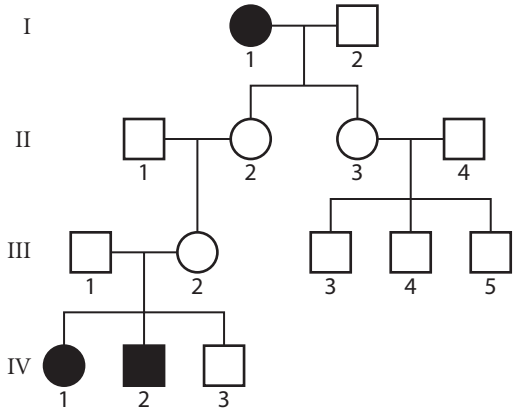


Figure 17.30 A pedigree showing the inheritance of PKU, an autosomal recessive trait. Notice that affected children may have unaffected parents, and that heterozygotes are not affected by the trait. How do you know that individual III 1 is heterozygous?

Sex-Linked Traits

The pedigree in Figure 17.31 shows the inheritance of hemophilia in the European royal families. Hemophilia is a condition that affects the body's ability to produce the proteins involved in blood clotting. People with hemophilia can suffer serious blood loss from simple cuts and bruises. The pedigree shows that males are at a much greater risk of being affected by hemophilia than females. It also shows that women who do not have the condition can pass on the trait to their children. From this information, you can deduce that hemophilia is an X-linked recessive trait.

You have already studied colour blindness, which is an X-linked recessive trait. Duchenne muscular dystrophy is another X-linked recessive trait. People

BiologyFile

FYI

People with PKU must not eat diet foods or drink diet soft drinks that contain the sugar substitute aspartame. Aspartame contains phenylalanine.

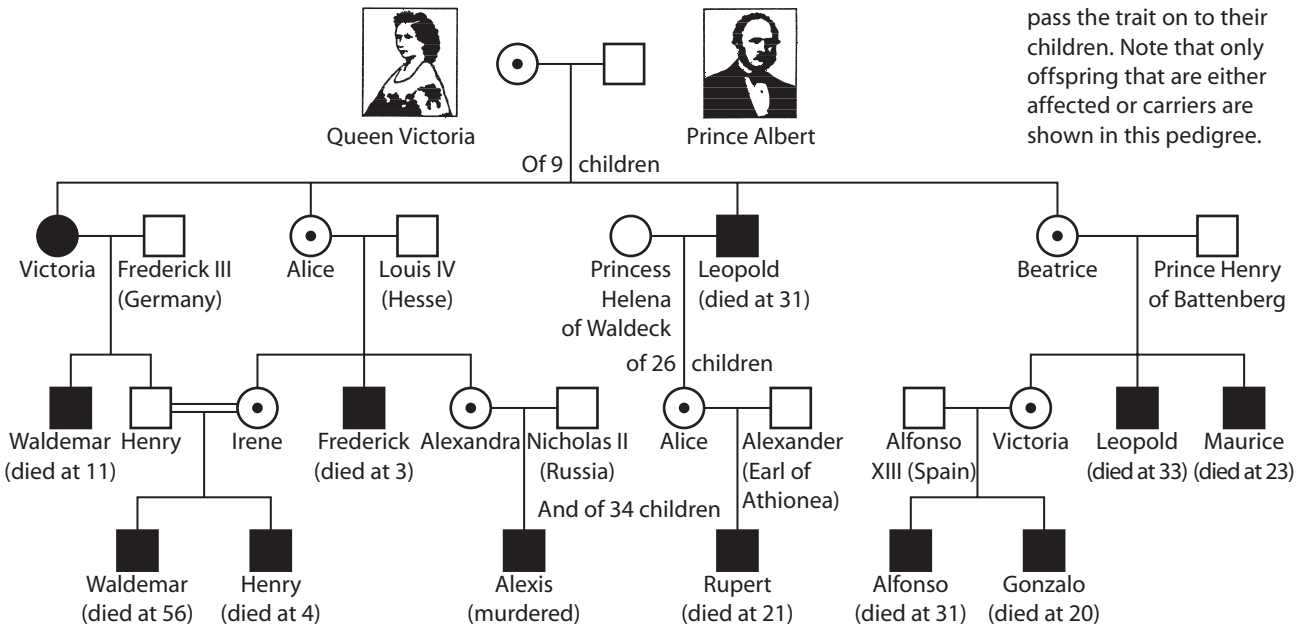


Figure 17.31 Pedigree showing the inheritance of hemophilia, an X-linked recessive trait, in the European Royal families. Males are much more likely to have hemophilia than females. Females who are heterozygous for the condition (carriers) can pass the trait on to their children. Note that only offspring that are either affected or carriers are shown in this pedigree.

BiologyFile

FYI

The first human gene to be mapped was the gene for red-green colour blindness. This gene was mapped to the X chromosome in 1911. Since then, more than 300 human genetic disorders have been mapped to the X chromosome.

with this disorder cannot manufacture the muscle protein dystrophin. As a result, their muscle tissues weaken and degenerate over time. The symptoms usually appear within the first three to five years of life and become increasingly severe with age. Because the muscles of the heart and respiratory system are also affected, the life expectancy of people with muscular dystrophy is only about 20 years.

Q32 How do pedigrees for autosomal recessive and X-linked recessive traits differ?

Q33 Can a female have hemophilia? Explain.

Human Genetic Analysis

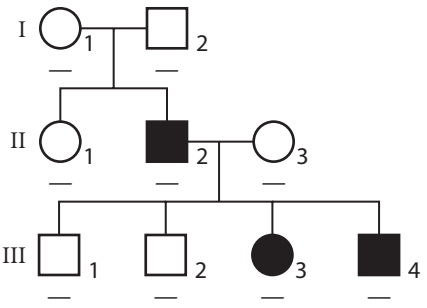
In 1902, an English physician named Sir Archibald Garrod (1857–1936) coined the phrase "inborn errors in metabolism" to describe four conditions that he realized were inherited: alkaptonuria, albinism, cystinuria, and pentosuria. Garrod also realized that these conditions were caused by the absence of specific enzymes. A few years later, he wrote a book called *Inborn Errors in Metabolism*. Although the field of genetics was in its infancy, Garrod saw patterns of inheritance in the four conditions.

Since Garrod's time, many more genetic conditions have been identified and mapped to specific chromosomes. Table 17.3 lists a few examples. Chromosome mapping helps geneticists develop new ways to identify people

Sample Problem

Problem

The following pedigree shows the inheritance of cystic fibrosis, an autosomal recessive trait, in a family. Identify the genotypes of each family member represented in the pedigree.



What Is Required?

The genotype of each individual in the pedigree.

What Is Given?

The pedigree is given.

Plan Your Strategy

Step 1 Look for an individual with a phenotype that differs from the corresponding phenotype in both parents. This phenotype must result from a homozygous recessive phenotype.

Step 2 Write the symbol for the dominant allele below every individual who does not show the trait.

Step 3 Both parents of the individuals showing the trait must have at least one recessive allele. All the children of a person showing the trait had to receive one recessive allele from this parent.

Act on Your Strategy

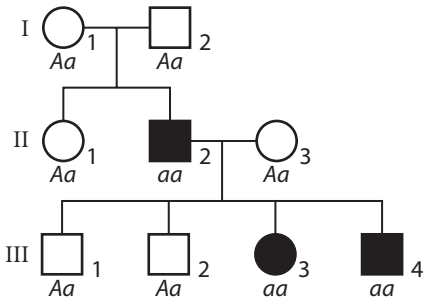
Step 1 Individual II 2 has a different phenotype. Write a homozygous recessive genotype (*aa*) below the symbol for all the individuals who show the trait (II 2, III 3, and III 4).

Step 2 Write the symbol for the one dominant allele (*A*) below all open symbols.

Step 3 Write "*a*" beside "*A*" for I 1, I 2, and II 3.

Step 4 Write "*a*" beside "*A*" for III 1 and III 2.

Step 5 You cannot determine whether II 1 is heterozygous or homozygous dominant.

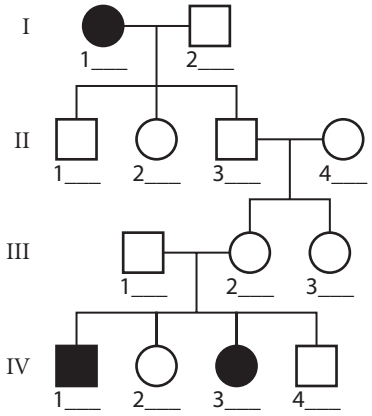


Check Your Solution

Upon checking the pedigree, all genotypes are correct.

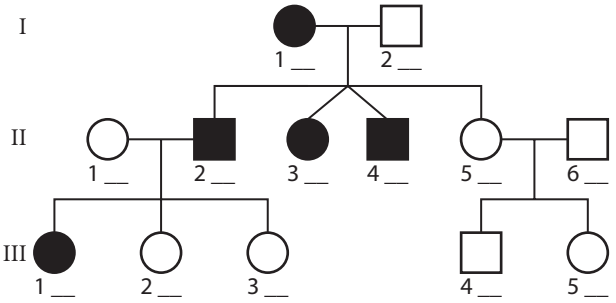
Practice Problems

18. A curved "hitchhiker's thumb" is recessive to a straight thumb. The following pedigree traces the presence of hitchhiker's thumb in a family. Identify the phenotypes and genotypes of all the people shown in the pedigree. Whose genotypes can you not be certain of?

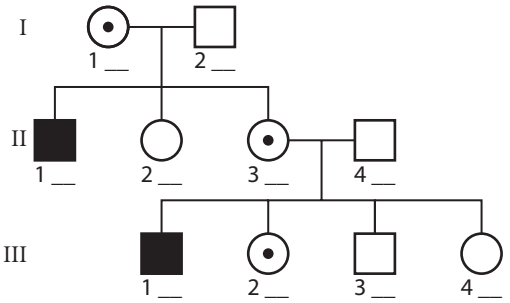


19. In certain families in Norway, woolly hair (hair that looks like sheep's wool) is passed down through the generations. In order for children to have this trait, at least one of their parents must have woolly hair. How is this trait most likely inherited? Draw a pedigree for a family where one of three children and both parents have woolly hair. Identify the genotypes and phenotypes for each individual in the family. Whose genotype can you not be certain of?

20. This pedigree traces tongue rolling in a family. The ability to roll your tongue is controlled by a dominant allele; people with the recessive allele cannot roll their tongue. Identify the phenotypes and genotypes of all the people shown in this pedigree.



21. Duchenne muscular dystrophy is an X-linked recessive trait. The following pedigree shows the occurrence of this disorder in an extended family. Provide the phenotypes and genotypes of all the individuals in the pedigree.



Thought Lab 17.2 Creating a Pedigree

Target Skills

Designing a plan to collect data in order to demonstrate human inheritance

Drawing and **interpreting** pedigree charts from human inheritance patterns

Although most characteristics in humans are influenced by more than one gene, some are controlled by single genes. Each of the following is a single-gene trait.

- shape of hairline peaked or smooth
- clasped hands left thumb on top or right thumb on top
- hair on middle segment of fingers or no hair on middle segment of fingers
- tongue rolling ability or inability to roll tongue
- thumb straight or bent back
- earlobe detached or attached

Procedure

1. Work with a partner. Choose a single-gene human trait. Then choose a family to interview. This can be your family or your partner's family, or another family if you know a large number of family members.
2. Interview family members, including grandparents, aunts, uncles, and cousins. Find out which phenotype each person exhibits for your chosen trait.
3. Using the symbols in Figure 17.28 create a pedigree for the trait.

Analysis

1. From the pedigree, determine whether the trait is due to a dominant or recessive allele.
2. Fill in as much of each genotype as you can with the data you have.
3. Do all the data fit the patterns you might expect? If not, what factors could explain the difference?

Table 17.3 Examples of Single-Gene Conditions Mapped to Chromosomes

Chromosome	Condition	Description
4	Huntington's disease	degenerative neurological disease that results in loss of muscle control and dementia
7	cystic fibrosis	condition that causes thick, sticky mucus to build up in the lungs, making breathing difficult and leading to infection; also blocks the pancreas, stopping digestive enzymes from reaching the intestines
12	phenylketonuria (PKU)	condition that prevents breakdown of phenylalanine, causing an accumulation of phenylpyruvic acid, which results in developmental delays in cognitive function
13	retinoblastoma	tumour development on the retina of young children, which is usually fatal if not treated
15	Marfan syndrome	disorder of the connective tissues, causing weakness in the heart, blood vessels, and skeleton, as well as very long limbs
	Prader-Willi syndrome	developmental disorder, causing decreased muscle tone, short stature, and an insatiable appetite; can lead to life-threatening obesity
16	alpha thalassemia	condition in which defective hemoglobin binds oxygen poorly
18	Nieman-Pick disease	brain and nervous system impairment due to accumulation of lysosomes filled with cholesterol
19	maple syrup disease	inability to break down three amino acids, causing an accumulation of by-products and nerve degeneration; usually fatal if untreated
20	severe combined immunodeficiency disease (SCID)	a deficiency in one enzyme, resulting in minimal immune response and susceptibility to all diseases; bone marrow transplant used to replace immune system

who are at risk of developing particular genetic conditions or of passing these conditions to their children. This process is called **genetic screening**. *Drosophila* research has been very helpful in this work, since many human genetic disorders have counterparts in the fruit fly.

Conditions such as alkaptonuria (which causes the affected individual to produce black urine) and albinism (which is a lack of the skin pigment melanin) are rare, but not life threatening. As you have seen, other genetic conditions can cause severe—even fatal—health problems. For this reason, geneticists and medical scientists around the world invest a great deal of effort and money into researching new methods of genetic diagnosis and screening. Every advance brings new questions, however.

If you have the allele for Huntington's disease, for example, would you want to know that you will eventually develop this degenerative neurological disease? If you could find out whether you are a carrier for a disease such as cystic fibrosis, would it make a difference in your decision about having a family? If you were expecting a baby, would you want to know that the baby had a serious inherited condition?

Many people who grapple with questions such as these seek advice from **genetic counsellors**. Genetic counsellors can estimate the risk of inheriting a particular genetic condition. As well, they can explain the symptoms of genetic conditions and the available treatments, provide other information, and, equally importantly, give emotional support.

Section 17.3 Summary

- Although selective breeding has been practiced for centuries, an understanding of Mendelian genetics enables modern scientists to be very selective in the development of new plant and animal breeds.
- A pedigree is a key tool for geneticists who study the inheritance of human traits.
- Analysis of a pedigree indicates whether a trait is autosomal dominant, autosomal recessive, or sex-linked.
- A pedigree can provide information about the genotypes and phenotypes of previous generations, and it can be used to predict the genotypes and phenotypes of future offspring.
- Other methods of genetic screening and diagnosis can also provide information about human genotypes.

Thought Lab 17.3 Analyzing Pedigrees

Target Skills

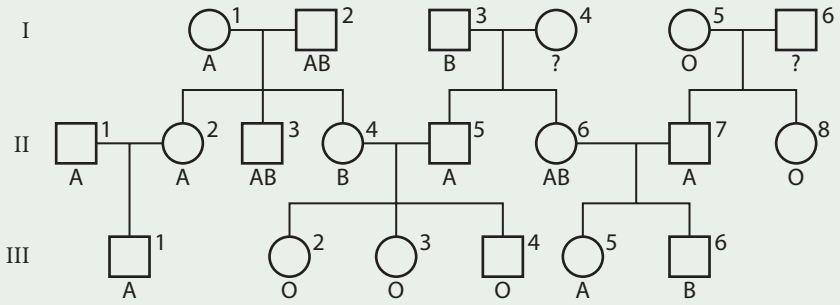
Drawing and **interpreting** pedigree charts from human inheritance patterns

Procedure

You have seen a pedigree can be used to trace the inheritance pattern of a single-allele trait. A pedigree can also be used to study multiple-allele traits. In the pedigree below, different blood types are identified by the letters A, B, AB, and O. Examine the pedigree, and then answer the questions that follow.

Analyze

1. Neither individual I 4 or I 6 has ever had their blood tested. What are their blood types?



2. Write the genotypes of as many individuals as you can. Whose genotypes can you not be sure of? Explain.

3. Individual III 3 marries a man with blood type AB, and they have four children. Will any of these children have blood type O? Explain.

4. Individuals II 1 and II 2 have a second child. After you see the child's blood-test results, you know that both parents have the genotype I^Ai. What blood type is the child?

5. Using examples from the pedigree, describe some of the limitations that apply to human genetic analysis. In what ways could these limitations affect genetic research?

6. Blood typing has often been used as an aid in paternity disputes. Can a blood test ever prove that a man is definitely the father of a particular child? Explain.

Section 17.3 Review

1. Give two examples of plants that have been developed through selective breeding techniques.

2. In what ways do the methods of researchers studying human genetics differ from the methods of researchers studying *Drosophila*? Explain.

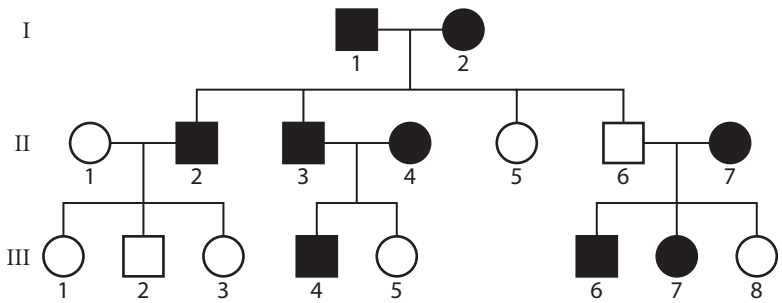
3. Examine the following pedigree.

a) What can you deduce about the pattern of inheritance of the trait?

b) Give one example of a human genetic condition that is passed on in this way.

c) What is the genotype of individual I 1?

d) How do the phenotypes of individuals III 1, III 2, and III 3 differ from the predicted ratios? What might account for this difference?



4. You have discovered a new autosomal recessive genetic condition. Individuals with this condition do not survive long after birth, and so do not have children of their own. Will the trait be eliminated from the population over time? Explain.

5. You suspect that you may be a carrier for a serious genetic condition, and you decide to consult a genetic counsellor. What three characteristics would you want the genetic counsellor to have?

Biobanks

Stored within the cellular fabric of your body tissues and fluids are the biochemical "bytes" that define and comprise your genetic identity. Do your genes belong to you and to no one else?

The Canadian Biotechnology Advisory Committee defines a biobank as "a collection of physical specimens [from a wide sampling of individuals] from which DNA can be derived and the data that can be derived from these DNA samples." A primary goal of biobanking is to "open source" millions of high-quality samples collected from a diverse group of world citizens for the medical benefit of humanity.

Some Key Questions

Human biological materials are routinely obtained during diagnosis and surgery. When diseased tissue is removed, part of the specimen is frequently retained for future clinical, research, or legal purposes. As well, volunteers often donate their bodies, organs, or blood for educational purposes, transplantation, or research. Unlike other information that is used for health purposes, however, genetic information is not just *about* us. Because of its uniqueness, it *is* us. In the future, who will have access to this information and what safeguards will be required? Will this information be available to insurance companies, employers, law enforcement agencies, or marketers of products and services?

Genes hold information not only about the donor, but also about other family members. While welcoming the potential health benefits of genetic research, many people have reservations about possible infringements on their privacy and human rights. One purpose of establishing large-scale human genetic research databases is to create research resources for the future. This raises other questions. Should individuals be contacted for each new use of their data? Should they be financially compensated if the use of their data results in the production of a life-saving drug?

Canada's Plan

Population biobanks are already established or being planned in Sweden, Iceland, Estonia, Tonga, the United Kingdom, and several other countries. Canada is developing a comprehensive position on biobanking, which will be followed by appropriate legislation to outline, delimit, and regulate the activities of institutions or companies that conduct biobank research.

In a background paper called *Survey of National Approaches to the Development of Population Biobanks*, prepared for the Canadian Biotechnology Advisory Committee in 2003, nine major challenges were put forward for Canada to consider when developing biobanking policies. These challenges are listed below.

Biobanking challenge for Canadians	Example of an associated question or issue
consultation	What steps are necessary to obtain and maintain the public trust?
recruitment	Which individuals, from which segments of the population, will be sampled?
consent	How will the approval of individuals, and Canadian society as a whole, be obtained?
governance	Who's in charge?
commercialization	What rights of ownership do sampled individuals give up—and to whom?
privacy	How will privacy be ensured, and who will be accountable?
communication of research results	Should individuals have access to their personal results or only to those of the aggregate (that is, the population)?
contribution to the welfare of the population	What constitutes a benefit in terms of a population, and how is this to be measured?
contribution to the welfare of humanity	How will all humanity share in and have access to the benefits of biobank research?

• • •

1. Find out how one of the countries that has already established biobanking addresses the moral, social, and legal issues.
2. Biobanking in Canada will be a reality in your lifetime. What opinions do you have and how can you voice them, now and in the future? What questions do you need answered to help you inform and clarify your opinions? What are the possible consequences of "passing the buck" where this particular issue is concerned?

Gregor Mendel was the first person to apply statistical methods to the study of inheritance. Mendel observed that heterozygotes do not express recessive traits, but can pass on these traits to their offspring.

Mendel's law of segregation states that all individuals have two copies of each factor (gene). These copies segregate randomly during gamete formation, and each gamete receives one copy of every factor.

Mendel's law of independent assortment states that the two alleles for one gene assort independently of the alleles for other genes during gamete formation. Parental genotypes can be inferred from the ratio of phenotypes among offspring.

Dominant traits mask recessive traits in heterozygotes. When alleles are co-dominant or incompletely dominant, heterozygotes have a different phenotype from both the homozygous dominant and the homozygous recessive.

Sutton observed that the pattern of Mendelian inheritance follows the movement of chromosomes during meiosis. He proposed the chromosome theory of inheritance. Morgan found that his experimental results could be explained if genes were arranged in a linear manner along chromosomes. He proposed the gene-chromosome theory.

Linked genes do not assort independently. Instead, the probability of recombination among linked genes increases with the distance that separates the two genes. Phenotype ratios can be used to calculate the map distance between linked genes.

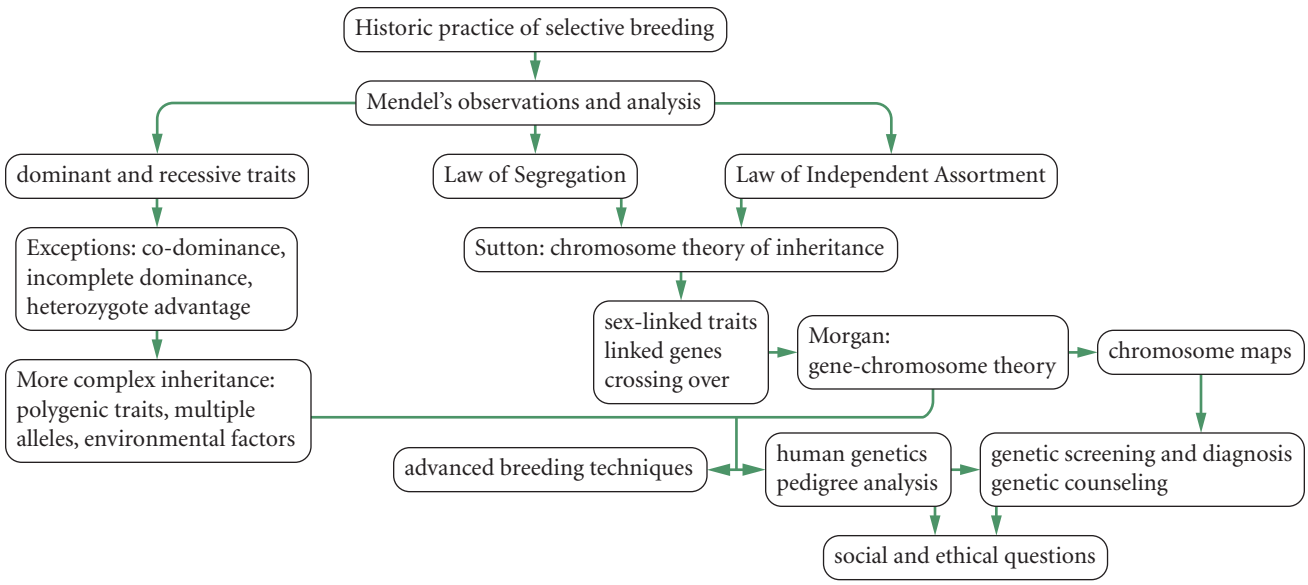
Not all traits follow the same patterns of inheritance. Polygenetic traits, and the presence of multiple alleles all result in genotype ratios that differ from Mendelian ratios in offspring.

Sex-linked traits have distinct patterns of inheritance. Recessive traits that are carried on the X chromosome are expressed far more frequently in men than in women. Environmental factors also can influence the expression of certain traits.

An understanding of the mechanisms and patterns of inheritance allows humans to develop new breeds of plants and animals that have desired traits.

Human inheritance follows the same principles as inheritance in other organisms. Different approaches are required to study human genetics, however. A pedigree is a tool that is used to trace patterns of inheritance of particular human traits. Pedigrees show distinct patterns for the inheritance of autosomal dominant traits, autosomal recessive traits, and sex-linked traits.

Chapter 17 Graphic Organizer



Understanding Concepts

1. Give three reasons that pea plants were an excellent choice for Mendel's research.

2. What is Mendel's law of segregation?

3. In humans, drooping eyelids are dominant to non-drooping eyelids.
- **a)** What are the two possible genotypes that a person with drooping eyelids might have?
- **b)** A man who is heterozygous for both alleles marries a woman with non-drooping eyelids. Use a Punnett square to show the expected genotypes and phenotypes of their children.
- **c)** A man with non-drooping eyelids and a woman with drooping eyelids have three children, all with non-drooping eyelids. How can you explain this outcome?

4. In humans, the gene for albinism is recessive to the gene for normal pigmentation. Your friends Milan and Aila both have normal pigmentation. They have one child who is an albino and are expecting a second child. Milan tells you, "We knew that our chance of having one albino child was 1:4. This means our chance of having *two* albino children is only 1:16, so we can be very sure that our next baby will not be albino." How would you respond? Using a specific example, explain to Milan whether or not his reasoning is correct.

5. A cross of true-breeding purple-flowered and true-breeding white-flowered plants results in F_1 plants that are all lavender (light purple). A cross of two F_1 plants results in an F_2 generation with the following numbers of phenotypes: 28 purple, 52 lavender, and 19 white.
- **a)** What were the genotypes of the F_1 generation? Explain.
- **b)** Use a Punnett square to show the predicted phenotypes and genotypes of the F_2 generation.
- **c)** Describe the type of inheritance that this cross reveals.

6. What is a test cross, and when is it used?

7. In what way does the chromosome theory of inheritance require a revision to Mendel's law of independent assortment?

8. A male mouse with a grey coat is mated with an albino female. Their six offspring all have grey fur. The albino female is then mated with a second grey male mouse. Four of the seven offspring in this litter are albino.
- **a)** What are the probable genotypes of the three parent mice?
- **b)** A male from the first litter is mated with a grey female from the second litter. What is the expected ratio of phenotypes among the offspring?

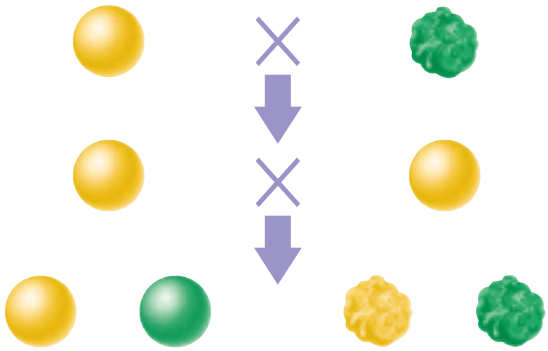
9. A man with type B blood marries a woman with type AB blood. What blood types would you expect to find among their children? What would tell you that the man is heterozygous for type B blood?

10. A woman with type AB blood has a child with the same blood type. What are the possible genotypes of the father?

11. Explain the significance of the Barr body in the expression of sex-linked traits.

12. A breeder of show horses finds that one of his stallions has a genetic defect that affects the production of sperm. The gene associated with this trait is located on the Y chromosome. What is the possibility that the stallion's female offspring could pass on this trait to their sons? Explain.

13. The following diagram shows the results of two crosses. Explain the results and the genetic principle that is illustrated.



Applying Concepts

14. In foxes, a pair of alleles, *P* and *p*, interact as follows: *PP* is lethal, usually during the embryonic stage; *Pp* produces platinum-coloured fur; and *pp* produces silver foxes. Could a fox breeder establish a true-breeding variety of platinum foxes? Explain.

15. Rudy and Sinead are expecting a baby. They have normal vision, but both of their fathers are colour blind. Determine the chance that the baby will be
- **a)** a colour-blind girl
- **b)** a boy with normal vision

16. Fruit flies can have normal wings or stunted wings. In an experiment, you mate several normal-winged females with a male that has stunted wings. In the F_1 generation, only the males have stunted wings.
- **a)** What can you conclude from this experiment?

b) Design an experiment to demonstrate whether or not females in the F_1 generation can also have stunted wings.

17. Imagine that the first dihybrid crosses Mendel performed had involved traits controlled by closely linked genes.

a) How would Mendel's results have differed from the results of a dihybrid cross involving non-linked genes?

b) What hypothesis might Mendel have developed to explain his results?

c) What experiment could Mendel have performed to test this hypothesis? What would he have observed?

18. Among some breeds of cattle, there is a trait called *polled*. Polled individuals do not develop horns. Suppose that a farmer breeds the same two polled individuals several times and five calves are born. Three of these calves are polled, and two are not. Is this an example of dominant or recessive inheritance? Explain.

19. A farmer wants to breed a variety of taller corn.

a) How can the farmer use variation in the height of the current corn plants to produce taller corn plants?

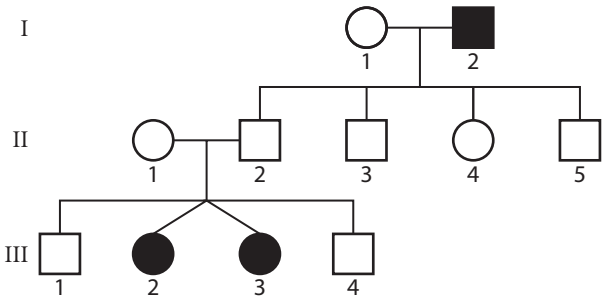
b) Will the farmer's work be most effective if height in corn plants is determined by polygenic inheritance, multiple alleles, or co-dominant alleles? Explain.

c) The farmer finds that many of the tallest corn plants are also very susceptible to a particular disease. How could the farmer design an experiment to find out if the genes for height are linked to the genes for resistance to the disease?

d) If these genes are linked, what steps could the farmer take to create a breed of corn that is taller and more disease-resistant than the current corn crop?

20. Explain why genes that are more than 50 map units apart on a single chromosome may behave as though they are on different chromosomes.

21. Osteogenesis imperfecta (OI), also known as brittle bone disease, results in extremely fragile bones that tend to break for no apparent reason. The following pedigree traces OI in a family. Based on the pedigree, what sort of inheritance pattern does OI display? Identify the phenotypes and genotypes of all the people shown in this pedigree. Whose genotype can you not be sure of?



Making Connections

22. Imagine that you are a genetic counsellor. Brian and Sarah come to see you. They are planning to start a family, but they want to know if their children are at risk for cystic fibrosis. Brian's sister has cystic fibrosis. Brian does not have the disease, nor do his parents.

a) What kind of questions do you think Brian and Sarah will ask?

b) How would you answer these questions?

c) What other information could you gather to help you define the risk?

23. Certain populations and cultural groups interest genetic researchers because they may have unique genetic characteristics. The study of these characteristics can contribute to the development of genetic tests and treatments that advance medical research. Who should have access to the genetic information of individuals from these populations and groups? Is this private information, or does it belong to all humanity? Discuss this question with a partner, and brainstorm your ideas. Prepare a presentation that outlines ideas on both sides.

24. People who are heterozygous for a recessive trait do not express the trait, but may pass it on to their children. Do we have a responsibility to inform our children of certain recessive traits they may have inherited from us? Present your thoughts in a short essay.

25. Many breeds of dogs are known for a high incidence of genetic disorders. German Shepherd and Saint Bernard dogs, for example, are predisposed to develop a crippling condition called hip dysplasia. Why are purebred dogs more at risk for such conditions than mixed breeds are? What advice would you give to dog breeders who want to maintain their dogs' purebred pedigrees, but also want their dogs to be as healthy as possible?