UNIT

6

Genetic Continuity

Unit Preview

In this Unit, you will discover

- how characteristics are inherited,
- how DNA can store and transmit hereditary information
- how the information encoded in DNA determines the features of an organism
- issues raised by genetic research and reproductive technology.

Unit Contents

High above the ground, within the coastal forest of Australia, you hear loud chatter and screeches. A flock of brightly coloured rainbow lorikeets gather in a treetop. Of the 58 species of parrots that live in Australia, each has plumage that is unique. No two species of parrot there have the same feather colour and pattern. Why do rainbow lorikeets have red, blue, and yellow feathers? Feather colour is a characteristic, or trait, that is inherited. This trait, along with many other traits, is passed from parents to offspring through reproduction. In the same way, you inherit many of your traits from your parents.

The genetic material carried in your cells determines such features as the colour of your skin and the texture of your hair. Genetic traits are passed down from one generation to the next through an unbroken family line. It was only about 150 years ago, however, that researchers began to understand the patterns of inheritance. Since then, the field of genetics has seen one breakthrough after another as scientists progressed from puzzling over the structure of DNA to creating entirely new forms of life.

In this Unit, you will learn how traits are inherited using examples from living organisms. Some traits have a simple inheritance pattern, while the inheritance of other traits may be very complex. You will see how hereditary information can be stored and transmitted by molecules, and how this information gives rise to the unique set of traits that make up each human individual. Finally, you will explore some of the recent discoveries and techniques that promise to transform medical science, industry, and even our definition of life itself.

How do individuals inherit characteristics from their parents?

CHAPTER 16

Genetics and Heredity

Reflecting Questions

- Why do offspring inherit certain characteristics from their parents but not others?
- How do events at the cellular level influence patterns of inheritance?
- Can the way in which certain traits will be inherited among members of a family be predicted?

For centuries people have realized that certain physical characteristics are passed from one generation to the next. They used this knowledge to produce crops and livestock with desired characteristics. They bred plants selectively to produce heartier and more nutritious crops — a prime example of this ingenuity is the development of varieties of wheat. They bred livestock to produce offspring with certain characteristics, such as stronger oxen or cows that could produce larger quantities of milk. How these characteristics were passed from parents to offspring, however, remained a mystery.

Have you ever traced your family history and found that you very closely resemble one of your distant relatives? Members of a family have many similarities in appearance, such as height, eye colour, and hair colour. But there is also variation in how characteristics are inherited by offspring. In other words, offspring do not look exactly like their parents. Why are some characteristics passed on to offspring, but not others? Why does the inheritance of some characteristics skip one or more generations? These questions remained unanswered until the late nineteenth century, when an Austrian monk performed a series of simple experiments and discovered how heredity worked.

In this chapter, you will learn how characteristics are passed from parents to their offspring. You will learn how to use different models to predict the genetic make-up and physical appearance of offspring, and how more recent research — including studies of chromosomes — expands on early theories of inheritance patterns. With this knowledge, you will be able to infer the genetic make-up of parents based on data collected about the characteristics of offspring. You will also study a number of genetic disorders in order to identify characteristic patterns of inheritance. These patterns of inheritance can be traced through pedigrees, or family histories. In this chapter, you will learn how to construct and interpret pedigrees to deduce the likelihood of a trait being carried and expressed within a family.

Why do members of a family show similarities in appearance?

Chapter Contents

16.1 Genetics of Inheritance

OUTCOMES

- Define and explain key terms relating to heredity and genetics.
- Use probability techniques to predict the outcome of various genetic crosses.
- Demonstrate an understanding of the concepts of dominance and segregation.

Every living organism is made up of many different **traits**, or distinguishing characteristics, that make it a unique individual. Certain traits in plants and animals may have qualities that people want to promote. For example, dog breeders have used the variations in traits of hair colour to produce the yellow, black, and brown colour variations of Labrador retrievers. Such variations are achieved through selective breeding of individuals. That is, only those individuals that show the desirable trait are permitted to reproduce. People began selectively breeding plants and animals thousands of years ago. They knew from observation and experience that certain traits could be passed from one generation to another. This transmission of traits is termed **heredity**, and the traits that are passed on are said to be **inherited**.

Figure 16.1 Selective breeding is used to develop special varieties of dog.

Genetics is the branch of biology dealing with the principles of variation and inheritance in animals and plants. The study of genetics gives us greater understanding of how we can determine the likelihood of inheriting certain traits and helps explain and predict patterns of inheritance in family lines. For much of human history, people did not understand how babies were conceived and how heredity worked. Clearly, there was some hereditary connection between parents and children, but the mechanisms were not readily apparent.

The Greek philosopher Hippocrates, for example, (460–377 B.C.E.) theorized that every part of the body was involved in the production of the "seeds" of the parents. These seeds then fused together to give rise to a new individual. In the eighteenth century, in contrast, some scientists believed that sperm contained pre-formed embryos and that the only female contribution to the next generation was the influences of the uterus in which the embryo developed.

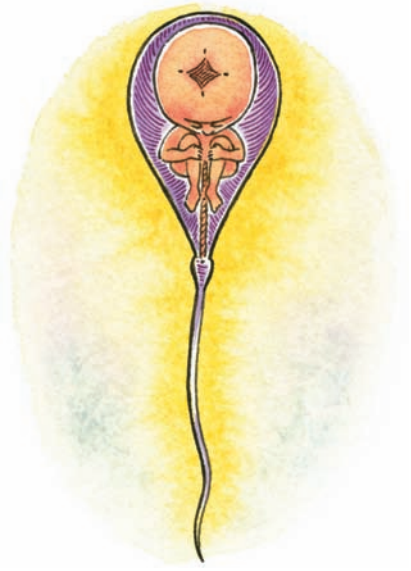


Figure 16.2 Just a few hundred years ago scientists proposed that sperm cells contained complete pre-formed embryos.

Figure 16.3 Gregor Mendel (1822–1884), an Austrian monk, studied garden pea plants as a means to explain the inheritance of characteristics.

Knowledge of the mechanisms controlling inheritance finally came as a result of careful experiments. This work was begun in 1853 by a monk in the monastery of St. Thomas in Brunn (now Brno, in the Czech Republic) named Gregor Mendel (see Figure 16.3). Before his work at the monastery, Mendel attended the University of Vienna, where his studies included mathematics and botany. This training became especially important during Mendel's study of heredity. He conducted a series of experiments on plants over an eight-year period (1853–1861). Following his experiments, Mendel published a paper outlining his conclusions.

Why did Mendel succeed in discovering the basis of inheritence when others before him had failed? There are three key points to any successful experiment in biology:

1. choosing the appropriate organism to study,
2. designing and performing the experiment correctly, and
3. analyzing the data properly.

Mendel conducted his research using the common pea plant (*Pisum sativum*), as shown in Figure 16.4. These plants were an excellent choice for four main reasons. The common pea plant was commercially available throughout Europe. It was easy to grow and matured quickly. Thirdly, the sexual organs of the plant are entirely enclosed in the flower. This means that pea plants self-pollinate, or fertilize eggs with pollen from the same flower, which allowed Mendel to control which plants reproduced. He introduced pollen from one flower to the pistil of another flower, cross-pollinating between plants to perform his experiments. Finally, different varieties of the common pea had different traits that could be observed easily from one generation to the next. Mendel examined seven different traits. Each trait had only two possible forms or **variations**. Mendel's decision to look at single traits helped him formulate his conclusions about heredity.

Figure 16.4 Common garden peas provided an excellent research organism for Mendel's experiments. These plants were easy to cultivate and had several traits that could be studied.

How did Mendel set up his experiment? The first thing he needed to do was to obtain purebred plants for the trait he wanted to study. A **purebred** organism is descended from ancestors of a distinct type, or breed. Purebred organisms in a given species or variety all share similar traits. For example, a certain variety of day lily may have a particular flower colour. This trait is inherited from previous generations, and results from a long period of selective breeding. Mendel produced purebred varieties of pea plants through selective breeding. He chose plants that were tall, for instance, and bred them together. These plants produced seeds, which Mendel planted and grew. Some of these plants grew tall and some were short. Mendel selected only the plants that grew tall and bred them together again. He continued to breed only

tall plants each generation until only tall offspring were produced. Mendel did the same for short plants, breeding only short plants together. In this way, he produced plants that were **true breeding**. That is, they only produced offspring that grew either tall or short. Mendel also produced true breeding pea plants for each form of the other six traits. Like plant height, the other traits were easily distinguished and could be classified into one of two categories.

BIO FACT

Although pea plants normally self-pollinate, plant breeders can also mate two different plants by hand. Cross-pollination is the method breeders use to transfer the pollen from one plant to another. In this way, two different true-breeding plants can be mated and the traits of the offspring studied.

Biology At Work

Plant Breeder

There is a pink and burgundy "Angel on My Shoulder." To my right is a salmon pink "Jazz Beat" and beyond that, a lemon yellow "Tuscany Lights." These are all varieties of day lilies that you might see at Henry Lorrain's *We're In The Hayfield Now Day Lily Gardens* near Orono, east of Toronto. Henry specializes in breeding new varieties of day lilies. He displays and sells them at his gardens, as well as supplying them to companies that resell them to gardeners.

Day lilies, a favourite with many gardeners, range in colour from red to pink to yellow to almost white. They are easily grown, tolerating a variety of temperatures, light conditions, and soils. They have a long growing season and, though their blossoms last only one day, a new crop of flowers bursts forth every morning.

Day lily breeders, Henry Lorrain and Douglas Lycett

Lilies of the Field

It was Henry Lorrain's partner, Douglas Lycett, who inspired Henry's interest in day lilies. Douglas Lycett died in 1998, but Henry continues his work. Using the method called hand pollination, Henry takes pollen from the stamen of one variety of day lily and applies it to the pistil of another. His aim might be, for example, to get a gold edge on a purple day lily or to produce a hardier day lily with prettier blossoms. Once Henry has pollinated the flowers, he waits for them to produce seeds. The following spring, he and his staff plant these seeds in seed trays and, when the ground is warm enough, they plant them outdoors. The plants do not bloom until the following year. "There's lots of variety in how the flowers look," Henry says. "Some are awful but others are spectacular." The plants with the spectacular flowers are the ones Henry chooses to propagate and eventually sell.

The goal of plant breeders like Henry Lorrain is to alter plants genetically to suit human needs and preferences. This can include developing anything from a gold-edged purple day lily, to oats that are more nutritious for horses, to a variety of potatoes that resist diseases (such as potato blight). Some plant breeders, like Henry, have their own companies. Some work for companies that supply seeds and bulbs to farmers and gardeners. Some do research and teach at universities. A number of Canadian plant breeders work for federal and provincial departments of agriculture, where they carry out research. They inform farmers and gardeners of new varieties, telling them which varieties are likely to grow best in certain conditions. They develop and enforce standards for the production and sale of seeds and other plant products. Some plant breeders work for international agencies, where they help to improve living standards in developing countries.

Career Tips

1. Plant breeding draws on many sciences including agriculture, biology, horticulture, botany, genetics, and plant biochemistry. Studying one or more of these sciences could lead you into a career in plant breeding. Some universities also offer programs specifically geared to plant breeding.
2. Find out more about one of the following: (a) Charles Saunders and Marquis wheat; (b) Baldur Stefansson and canola; (c) J. Patricia White and the Silken Laumann Rose.
3. Genetically modified foods have been both promoted and opposed for several years. Research this issue. Use what you learn as the basis for a pro and con chart, class debate, or letter to a company president.

Mendel's First Experiment: A Monohybrid Cross

Once Mendel had his purebred plants, he designated them the parent generation, or **P generation**. Then he crossed a true-breeding tall pea plant with a true-breeding short pea plant. The offspring from this cross were the first **filial generation**, or **F_1 generation**. Mendel called the F_1 generation **hybrid** plants to indicate they were the result of a cross between two different purebred plants. This is called a **monohybrid** cross, because only one trait, plant height, was involved.

What happened when Mendel planted the seeds of the F_1 generation? According to the theory of blending inheritance, he should have obtained plants of medium height. What Mendel observed, however, was that 100% of the plants in the F_1 generation were tall. (See Figure 16.5). This led Mendel to conclude that the trait for tall plants must be dominant, and the trait for short plants must be recessive. A **dominant** trait is a characteristic that is always expressed, or always appears, in an individual. A **recessive** trait is a characteristic that is latent (present but inactive) and is therefore not usually expressed in an individual. (A recessive trait may be expressed if it is the only trait present, as Mendel observed in his short-growing plants.) In this case, the trait for tall pea plants was dominant over the trait for short plants. All the pea plants grew tall if they possessed the dominant trait for size — tall.

Mendel conducted this experiment many times using true-breeding plants for each of the seven traits he had chosen to study. He obtained the same results every time: one trait was dominant over the other. Mendel concluded that heredity was definitely not just a blending of traits. He also concluded that when plants with two contrasting traits are crossed, one trait is always dominant over the other. This led him to formulate the **principle of dominance**: when individuals with contrasting traits are crossed, the offspring will express only the dominant trait. That is, if a purebred tall pea plant is crossed with a purebred short plant, all the offspring will be tall.

Law of Segregation

The next experiment Mendel conducted involved breeding the F_1 generation. He allowed the hybrid tall plants of the F_1 generation to self-pollinate. This produced the second filial generation, or **F_2 generation**. Figure 16.5 shows the traits that Mendel observed in the F_2 generation.

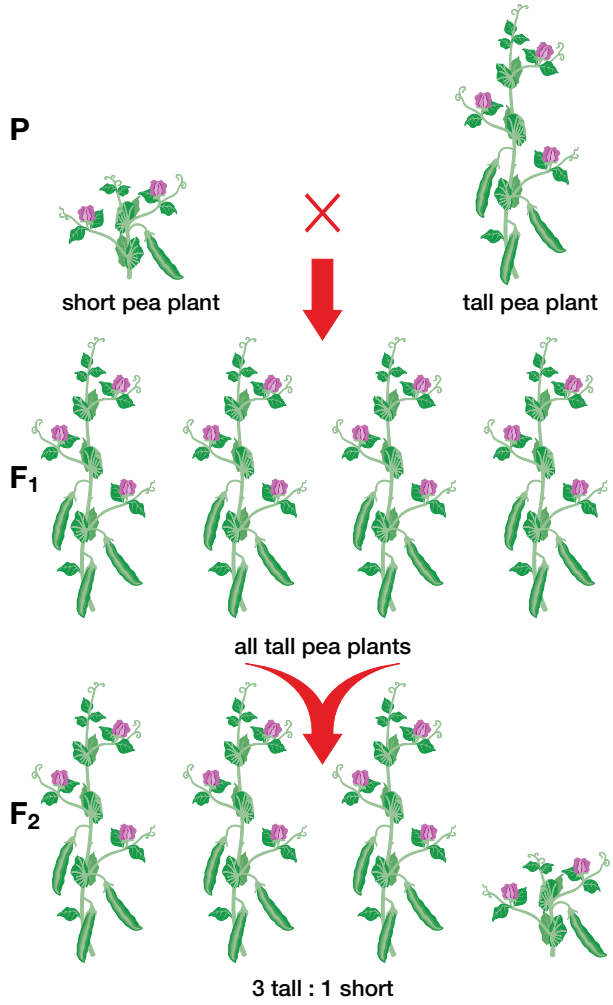


Figure 16.5 Mendel crossed a purebred tall pea plant and a purebred short pea plant. The resulting F_1 generation was all tall pea plants. Mendel then allowed plants of the F_1 generation to self-pollinate. In the F_2 generation, three quarters of the pea plants were tall and one quarter were short.

Three out of four plants in the F_2 generation were tall while one was short. Mendel repeated this experiment many times and examined all seven traits. He obtained the same results time after time. The F_2 generation resembled one parent from the P generation 75% of the time and the other parent from the P generation 25% of the time. This ratio of 3 : 1 is known as the **Mendelian ratio**. Why did this happen? Based on his observations of traits in the F_1 and F_2 generations, Mendel drew the following conclusions:

- Each parent in the F_1 generation starts with two hereditary "factors." One factor is dominant and the other is recessive.

- The factors separate in the parent. Only one factor from each parent is contributed to the offspring.
- Each offspring inherits one factor from each parent. If the dominant factor is present it will be expressed even if the recessive factor is also present.
- The recessive factor will be expressed if only recessive factors are present.

Mendel's results from the F_2 generation gave rise to his first law of heredity. The **law of segregation** states that inherited traits are determined by pairs of "factors." These factors segregate (separate) in the gametes, with one in each gamete. Recall from Chapter 14 how gametes are formed.

We know today that Mendel's "factors" were **genes**, the part of the chromosome that governs the expression of a particular trait. A gene can occur in alternate forms called **alleles**. When two alleles are present, a dominant allele may prevent the expression of the recessive allele. Even though the dominant allele is expressed and the recessive allele is not, the recessive allele has not been altered physically and will pass unchanged in an individual's gametes to the next generation, where it may or may not be expressed.

Because traits are inherited as independent units, Mendel's factors of inheritance are sometimes referred to as **unit characters** and his theory as the **unit theory** of inheritance. In Mendel's first experiment, he started with two purebred plants, as shown in Figure 16.6. Using letters to represent the different alleles, a purebred tall pea plant will have two uppercase letters, TT. In other words, the plant is **homozygous** for tall (the two alleles are the same). The purebred short pea plant has two alleles for short, designated in lowercase letters as tt. It is said to be homozygous for short. What happened when Mendel crossed the two plants? Each parent contributed one allele to each offspring — one T from the tall plant and one t from the short plant. The product of the cross was a tall offspring with alleles T and t. A plant that has a dominant and a recessive allele is said to be **heterozygous** for tall (the two alleles are different). The tall allele is dominant and is therefore expressed. The short allele is recessive and, while present, will not be expressed. Study Figure 16.7 to learn how alleles were distributed among the offspring when plants of the F_1 generation reproduced. Recall that the recessive trait is expressed only when there is no dominant allele.

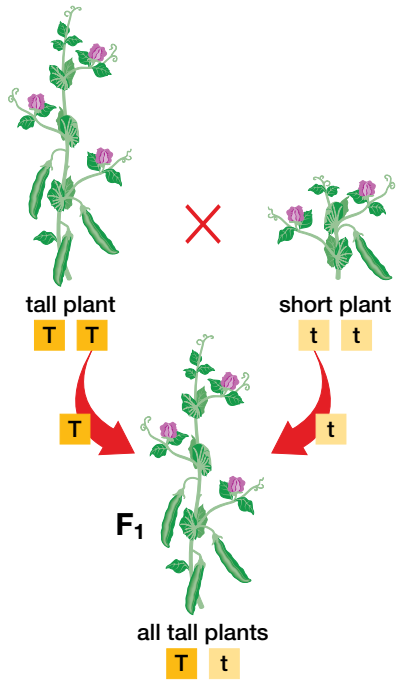


Figure 16.6 A cross of purebred short and tall pea plants illustrates Mendel's principle of dominance. Dominant alleles (T) are expressed in the F_1 generation and recessive alleles (t) are not expressed.

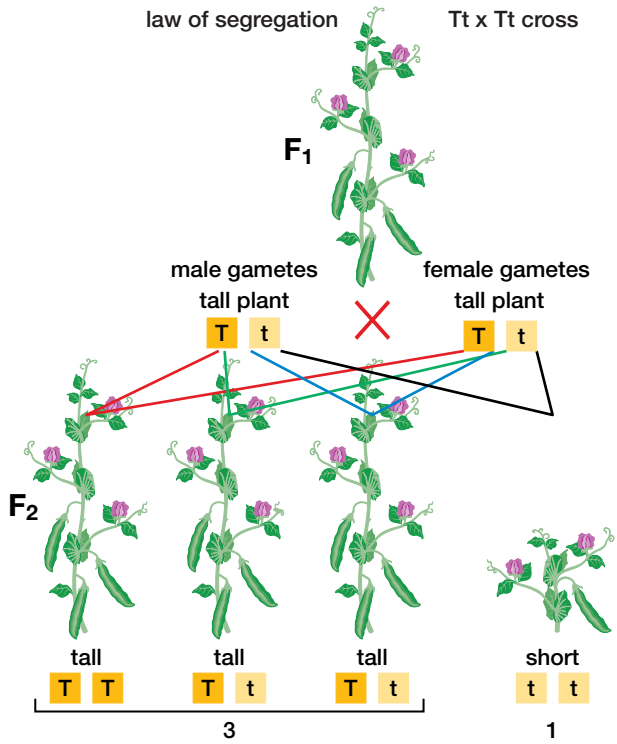


Figure 16.7 Individuals from the F_1 generation of pea plants produced a 3 : 1 ratio of tall plants to short. Mendel concluded that each parent plant had two factors and that these factors segregate during the production of gametes (eggs or sperm).

The Product Rule: Practice Problems

1. A coin is tossed in the air and allowed to land on the table. What is the probability that the coin will come up heads? Derive a simple formula that expresses the probability of this occurring.
2. Calculate the probability of throwing a pair of sixes when two dice are rolled.
3. What are the chances that a family of four children will have two boys and two girls? All girls? At least three boys?
4. In humans, albinism (lack of skin pigment) is due to a recessive gene. Suppose that two normally pigmented parents produce an albino child. What are the chances of their second child also being albino?

Probability and Genetics

If you flipped a coin into the air, what is the chance that it would land on the ground heads up? The coin could land tails up, too. In fact, there is an equal chance, or **probability**, of getting either a head or a tail. This probability can be expressed as a ratio — one head: one tail, or 50 : 50. This is the ratio you would expect because there is an equal chance for each outcome — heads or tails. What would be the possible outcomes if you flipped two coins at the same time? Complete the next MiniLab to find out.

The **product rule** states that the probability, or chance, that two or more independent events will occur together is the product of their individual probabilities of occurring alone. For example, what is the probability of flipping two coins and getting two heads at the same time? The chance of getting one head is $\frac{1}{2}$ or 50 : 50. The chance of getting the second head is also $\frac{1}{2}$ or 50 : 50. The chance of getting both heads at the same time is $\frac{1}{2} \times \frac{1}{2} = \frac{1}{4}$.

Figure 16.8 shows all possible outcomes of flipping two coins. Note that the chance of each result is not affected by the previous result. That is, getting

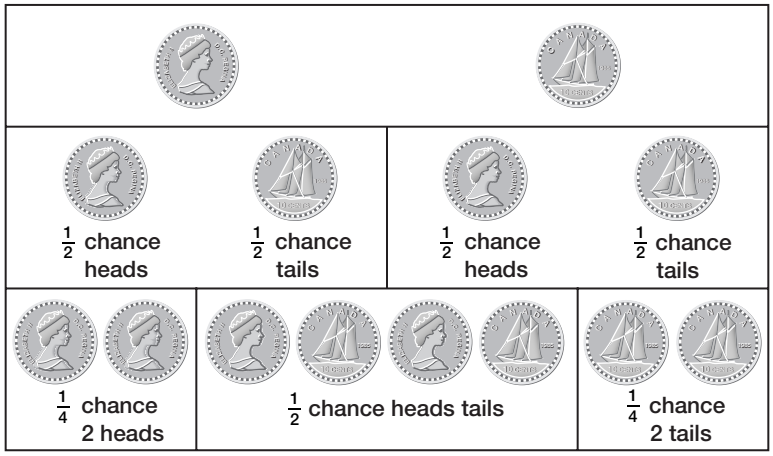


Figure 16.8 The product rule. There are two possible outcomes when tossing a coin in the air. To find the probability of several different combinations of outcome, you need to multiply the individual outcomes.

MINI LAB

Heads or Tails?

In this lab, you will investigate the probability of turning up heads or tails during the toss of two coins. Together with a partner, two coins, paper, and a pencil, flip one coin 20 times and record how many times you get heads and how many times you get tails. Tabulate the class results. Now both of you flip a coin at the same time and record how many times you get two heads, two tails or one of each. Do the toss 20 times. Use H for heads and T for tails. Tabulate the class results.

Analyze

1. What do the results of the first part of the experiment seem to show? How many times did you record heads? How many times did you record tails? What is the probability of either heads or tails being the result of each toss? Do the class results support this?
2. In the second part of the experiment, what are the chances of two heads turning up? Of two tails turning up? What is the chance that you will turn up one of each? Is it greater or less than the chance of turning up both heads or both tails? Do the class results support this?
3. How does this experiment help illustrate ratios and probabilities in inheritence?

heads in one toss does not make the next toss more or less likely to turn up heads.

The law of probability forms the basis for solving genetics problems. The two alleles for each parent represent probabilities. For example, in a cross of pea plants that are heterozygous for tall (Tt), it is probable that half of the gametes will contain the T allele and half will contain the t allele. A gamete with the T allele may combine with a gamete with either another T allele or a t allele. This happens entirely by chance. The probability of getting a particular combination of alleles in a given zygote depends on the genetic makeup of the parents.

Punnett Squares

What are the possible combinations of alleles in the offspring? The results can be organized easily in a **Punnett square** (see Figure 16.9). A Punnett square is used to calculate the probability of inheriting a particular trait. It is a simple method of illustrating all possible combinations of gametes from a given set of parents. All the possible gametes for one parent are listed across the top and all the possible gametes for the other parent are listed down the side of the square. Each box is then filled in by copying the row and column-head letters across or down the empty squares. This will give you a prediction of the outcome of a particular cross for a given set of alleles. Using the Punnett square, you can determine both the genotypes and phenotypes of the offspring of different crosses. The **genotype** is the genetic make-up of an organism. The **phenotype** is the appearance of the trait in an organism. For example, the genotype of the F_1 generation of pea plants is Tt but the phenotype is tall.

BIO FACT

The use of a Punnett square to determine the outcome of various crosses was first proposed by Reginald C. Punnett, an early twentieth century English geneticist who worked with traits of feather colour in chickens. He discovered certain fundamentals of genetics, including sex determination and traits that are linked, or specific, to each sex.

WEB LINK

www.mcgrawhill.ca/links/atlbiology

For an interactive exploration involving various crosses, go to the web site above, and click on **Electronic Learning Partner**.

Using the Punnett square, you can examine the genotypes and phenotypes of the $F_1 \times F_1$ cross. Recall that all of the F_1 generation of pea plants had the genotype Tt. The only possible gametes for both the mother and the father then are T and t. The Punnett square shows that this results in one genotype TT, two genotypes Tt, and one genotype tt. Expressed as a Mendelian ratio for genotype, this is 1 : 2 : 1. There is one homozygous dominant genotype, two heterozygous genotypes and one homozygous recessive genotype. The Punnett square also indicates the expected phenotype. Three of four offspring have the T allele and will be tall. Only one genotype, homozygous for the recessive characteristic, will result in a short pea plant. Expressed as a Mendelian ratio for phenotype, this is 3 : 1. Note that the phenotype of a Tt plant is indistinguishable from that of a TT plant. This is an example of **complete dominance**. Complete dominance is the type of inheritance in which both heterozygotes and dominant homozygotes have the same phenotype. Later you will learn about other forms of dominance.

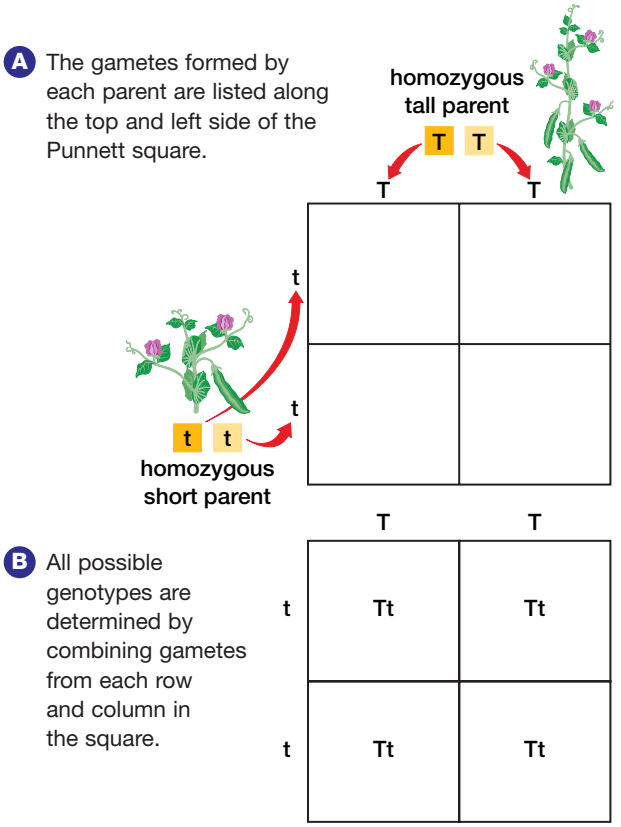


Figure 16.9 A Punnett square can be used to determine the outcome of crosses. According to Mendel, the alleles separate during gamete formation to produce T gamete and t gamete. Each gamete of one plant has the opportunity to pair with the gametes of the other plant. All possible pairings of alleles produce tall plants (Tt).

Sample Problem

Solving a Punnett Square

Tall pea plants are governed by the allele T while short pea plants are governed by the allele t. Suppose a heterozygous tall pea plant was crossed with a short pea plant. What are the genotypes of the parents? Identify the genotypes and phenotypes of the offspring of the F_1 generation.

What is required?

You are asked to find the genotype of each parent as well as the genotypes and phenotypes of the F_1 generation.

What is given?

You know the genotypes and phenotypes of each parent:

T represents the allele for tall plants

t represents the allele for short plants

For the heterozygous tall parent, the genotype is Tt

For the short parent (homozygous short) the genotype is tt (must be tt because of the law of dominance)

Plan your strategy

You are asked to make the following cross: Tt × tt

Act on your strategy

The genotypes of the parents are: Tt (heterozygous tall plant), and tt (homozygous short plant). Place the alleles of each parent along the columns and rows of the Punnett square and complete the possible crosses.

	T	t
t	Tt	tt
t	Tt	tt

The genotypes of the offspring are: two Tt and two tt. This is a genotypic ratio of 1 : 1.

These genotypes correspond to the following phenotypes:

Tt — heterozygous tall plant

tt — homozygous short plant

The corresponding phenotypic ratio is also 1 : 1.

Check your solution

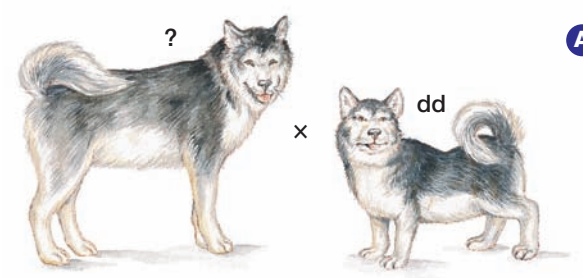
The parent genotypes are Tt and tt representing tall and short plants, respectively. The genotypes of the F_1 generation are Tt and tt, which represent the phenotypes tall and short plants, respectively.

Practice Problems

1. In pea plants, round peas are dominant over wrinkled peas. Use a Punnett square to predict the phenotypic and genotypic outcome of a cross between a plant homozygous for round peas (RR) and a plant homozygous for wrinkled peas (rr).
2. In tomatoes, red fruit (R) is dominant over yellow fruit (r). If a homozygous red fruit is crossed with a yellow fruit,
   - **(a)** What is the appearance of the F_1 generation?
   - **(b)** What are the genotypes of the F_2 generation if two plants from the F_1 generation are crossed?
   - **(c)** What are the phenotypes of the F_2 generation?
3. In cattle, horns (h) are recessive over hornlessness (H). If two homozygous cattle, one hornless and the other horned, are crossed, what are the genotypes and phenotypes of the first generation?

Determining Genotypes

How can you determine if a particular organism is a homozygous dominant or a heterozygous? You cannot deduce this information by looking at the phenotype alone. Recall that tall pea plants may be homozygous dominant (TT) or heterozygous (Tt). In order to determine the genotype of an individual, you must perform a **test cross**. A test cross involves crossing an individual of unknown genotype with a homozygous recessive individual. Figure 16.10 shows how test cross results are interpreted.



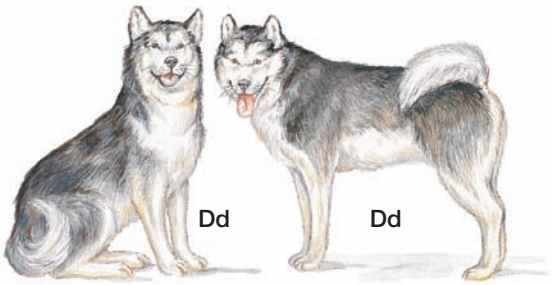
A The dog of unknown genotype could be heterozygous (Dd) or homozygous dominant (DD) for the trait.

B If the dog of unknown genotype is homozygous dominant (DD), all the offspring will be heterozygous (Dd) and will appear normal.

Homozygous × Homozygous

	DD	dd
	d	d
D	Dd	Dd
D	Dd	Dd

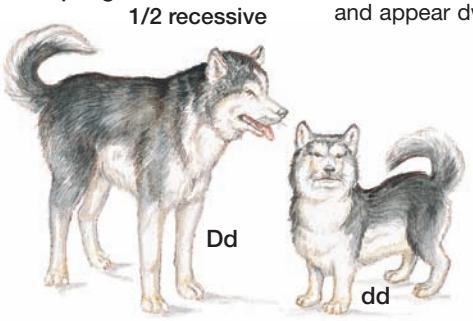
Offspring: all dominant



Heterozygous × Homozygous

	Dd	dd
	d	d
D	Dd	Dd
d	dd	dd

Offspring: 1/2 dominant
1/2 recessive



C If the dog of unknown genotype is heterozygous, half the offspring will be heterozygous (Dd) and appear normal. However, the other half of the offspring will be homozygous recessive and appear dwarf.

Figure 16.10 This test cross of Alaskan malamutes involves a dwarf dog that is homozygous recessive (dd) and a normal-size dog of unknown genotype (D?).

THINKING LAB

Identify the Genotypes

Background

It is often difficult to determine the exact genotypes of parents of various species when only their phenotypes are known. In this lab, you will deduce probable genotype(s) for parents based on a series of crosses. Carefully read each scenario and the outcome of each cross that is listed in the table, and then complete the questions.

You Try It

1. Suggest probable genotypes for the parents based on the results of each cross.
2. Give a brief explanation of how you determined the genotypes for each of the scenarios described in the table.
3. Identify and explain any evidence that enabled you to be reasonably sure of your answers.
4. Are there any answers of which you are unsure? Why or why not?
5. What additional information would you need in order to positively determine the genotypes of those that you were unsure about?
6. Identify the genetic principle (or principles) that enabled you to solve each scenario.

Scenario	Outcome (offspring)
A watermelon plant with striped fruit is crossed with a plant heterozygous for this characteristic.	All fruit are striped.
A black pig mates with a white pig.	In a litter of 16 piglets, 75% are black and 25% are white.
A yellow-haired female rat is mated with a black-haired male rat.	Of 99 offspring, 46 are black and 53 are yellow.
Two pea plants are crossed.	Plants with green pods and plants with yellow pods are produced in the ratio 3 : 1.
A tall green pea plant is crossed with another of its kind.	Of 18 plants produced, only one is short and yellow.

SECTION REVIEW

1. Define the following terms: genetics, heredity, trait, variation.

2. State the principle of segregation.

3. What important ratio appears in the F_2 generation of Mendel's monohybrid crosses? Explain why this ratio appears.

4. Differentiate between the following: (a) dominant and recessive; (b) gene and allele.

5. Define the "principle of dominance." Briefly describe the experimental results that led Mendel to this principle.

6. Why might a plant breeder be interested in knowing how certain traits are inherited?

7. Why was the pea plant a suitable organism for Mendel to conduct his experiments on heredity?

8. What is the purpose of a test cross? How is a test cross performed?

9. Using an illustration, explain the purpose of the Punnett square.

10. In pea plants, yellow peas are dominant over green peas. Use a Punnett square to predict the phenotypes and genotypes of the offspring from a cross between a plant heterozygous for yellow peas (Yy) and a plant homozygous for green peas (yy).

11. As a reward for being a good student, Mr. Singh gives you a rabbit named Bud. Bud has long hair. You introduce Bud to your other rabbit, Sarah, who has short hair. Later that school year, Bud and Sarah produce a litter of one long-haired and seven short-haired bunnies. If short hair is due to the dominant gene (S) and long hair to the recessive allele (s):

(a) What are the possible genotypes of Bud and Sarah?

(b) What phenotypic ratio would you expect in the offspring generation of a cross between Bud and Sarah? Create a Punnett square to show the results of the cross.

(c) How many of the eight bunnies were expected to be long-haired?

(d) The expected phenotypic ratio would be achieved in every case. Would you agree with this statement? Why?

12. Huntington disease is caused by a recessive allele. Linda has a father who died of Huntington disease. Linda's mother's father also died of this disease. Use the product rule to calculate the probability that Linda will develop the symptoms of Huntington disease.

13. In humans, albinism (the lack of skin pigmentation) is governed by a recessive allele (a) and normal pigmentation is governed by the dominant allele (A). Given this information, determine the genotypic and phenotypic ratios of the children expected from the following crosses:

(a) homozygous dominant x heterozygous

(b) heterozygous x homozygous recessive

(c) homozygous dominant x homozygous recessive

(d) heterozygous x heterozygous

(e) For each of the above crosses, state the percent likelihood of the first child being albino.

Determine the percent likelihood of having one normal child followed by two albino children in one family.

14. A hornless bull is crossed with three cows, A, B, and C. Cow A is horned and produces calf A′ which is also horned. Cow B is hornless and produces calf B′, which is horned. Cow C is horned and produces calf C′, which is hornless. Give the genotypes and phenotypes of all seven animals.

15. In mice, brown eyes (B) are dominant over blue eyes (b). In your lab, you perform a testcross by mating one brown-eyed male with one blue-eyed female, which produces a litter of eight brown-eyed offspring.

(a) What would you conclude about the genotype of the male mouse?

(b) Your lab partner says. "Not so fast. The testcross result could be just a matter of chance." Use the product rule to determine the possibility that you may be wrong.

16. Many characteristics are the result of single gene inheritance. For example, a widow's peak is inherited over a smooth hairline if the allele for the widow's peak is present. How do you think characteristics that are controlled by more than one gene might be expressed?

17. Can you think of any other organism that would have been a good candidate for Mendel to study the genetics of inheritance? Why did Mendel choose to study plants rather than animals or, indeed, humans?

16.2 Complex Inheritance Patterns

OUTCOMES

- **Explain how the concept of independent assortment influences the inheritance of more than one trait.**
- **Predict the outcome of dihybrid crosses.**
- **Demonstrate an understanding of the concepts of incomplete dominance, co-dominance, and multiple alleles.**

When Mendel performed his monohybrid crosses on pea plants, he was investigating one trait at a time. This method allowed him to determine the inheritance pattern of plant height, for instance, among generations. However, organisms are composed of many traits. The common pea, among other characteristics, has traits for colour, shape, and height. Fragrance in flowers also has a genetic basis and is particularly important in the cultivation of ornamentals, such as roses (see Figure 16.11). How are multiple traits (two or more) inherited? This was the question Mendel sought to answer in his next series of experiments.

Figure 16.11 Many varieties of plants, such as this rose, have been hybridized. Each may have a combination of traits including flower shape, colour, and fragrance.

Mendel's Second Experiment: A Dihybrid Cross

Mendel wanted to know if the inheritance of one characteristic influenced the inheritance of a different characteristic. For example, did pea shape influence pea colour? Mendel approached this question the same way he had approached the previous ones. First, he produced plants that were purebred for the traits he wanted to examine. Using the combination of the pea's shape and colour, for instance, he selectively bred pea plants until the offspring always had round, yellow seeds. These plants were homozygous dominant for both traits. He then bred plants that were homozygous recessive for both seed shape and colour. These individuals always had wrinkled, green seeds. Mendel then performed a **dihybrid cross**; that is, he crossed two pea plants that differed in two traits — pea shape and pea colour. The F_1 generation all had round, yellow seeds. Remember that round seed shape and yellow colour are dominant characteristics (see Figure 16.12). The offspring were therefore all heterozygous for the two traits. If R represents the dominant trait for pea shape and Y represents the dominant trait for pea colour, the genotype of the F_1 generation is RrYy. The phenotype is round and yellow seeds. What happens if you cross individuals of the F_1 generation? Figure 16.13 shows all possible genotypes from an F_1 cross.

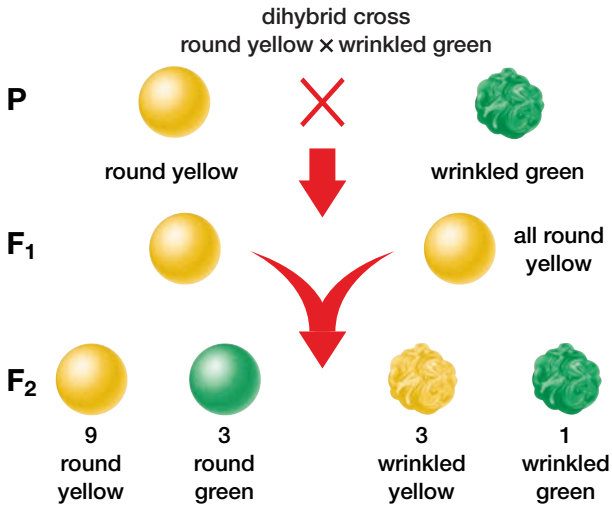
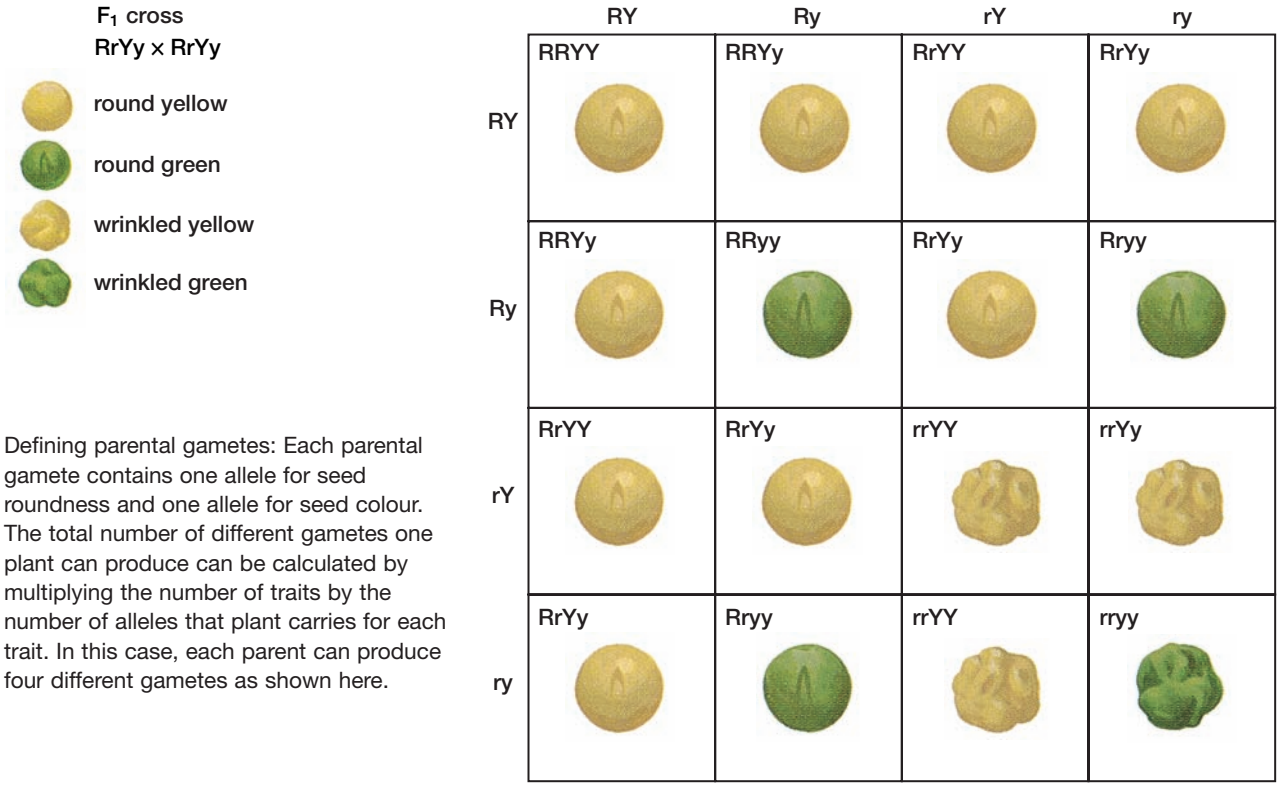


Figure 16.12 Results of Mendel's dihybrid crosses of pea plants. What traits were expressed in the F_1 and F_2 generations?



Defining parental gametes: Each parental gamete contains one allele for seed roundness and one allele for seed colour. The total number of different gametes one plant can produce can be calculated by multiplying the number of traits by the number of alleles that plant carries for each trait. In this case, each parent can produce four different gametes as shown here.

Figure 16.13 A dihybrid cross of the F_1 generation produced nine different genotypes. How many phenotypes resulted from this cross?

Mendel allowed the F_1 generation of his dihybrid cross to self-pollinate. Of 551 plants in the F_2 generation, Mendel observed the following traits:

- 320 round yellow
- 104 round green
- 101 wrinkled yellow
- 26 wrinkled green

These results represent a phenotypic ratio of 9 : 3 : 3 : 1. The F_2 generations of other dihybrid crosses for other traits showed a similar phenotypic ratio.

Law of Independent Assortment

Mendel realized that a ratio of 9 : 3 : 3 : 1 could be explained if the alleles from one trait were inherited independently of the alleles for another trait. This led Mendel to propose the **law of independent assortment**. This second law of inheritance states that the inheritance of alleles for one trait does not affect the inheritance of alleles for another trait. According to the law of independent assortment, different pairs of alleles are passed to the offspring independently of each other. This means that offspring may have new combinations of alleles that are not present in either parent. A pea plant's ability to produce white flowers instead of purple ones does not influence the same pea plant's ability to produce a round pea shape rather than a wrinkled pea shape.

Figure 16.13 illustrates how a Punnett square is used to determine the genotypes that result from a dihybrid cross. All the possible combinations of gametes for one parent are listed across the top, and all the possible combinations of gametes for the other parent are listed down the side.

WEB LINK

www.mcgrawhill.ca/links/atlbiology

The common fruit fly, *Drosophila melanogaster*, is often used by researchers to study the inheritance of traits. This fruit fly shows distinct phenotypes, such as eye colour and wing shape, which are inherited as simple dominants. Thus, genotypic and phenotypic ratios can be easily determined from crosses. A virtual computer lab may be used to investigate the inheritance of traits in this fly. To find out how to conduct crosses using virtual flies, go to the web site above, and click on **Web Links**. The virtual fly lab is recommended over conventional fly labs because there is no need to actually breed flies and wait for offspring to develop. Computer software allows you to create crosses and determine the outcome quickly and easily.

As in the case for a single trait, a test cross may also be used to determine the genotype of an individual for two traits. A two-trait test cross involves crossing an individual that shows the dominant phenotype for two traits with an individual that is homozygous recessive for the same two traits. The individual showing the dominant phenotype for both traits may be either heterozygous or homozygous dominant. A homozygous recessive individual is used in the cross because it provides the best chance of producing an offspring that is homozygous recessive for both traits.

For instance, if a pea plant is homozygous dominant for purple flower colour (PP) and round pea shape (RR), then the F_1 generation will all have the dominant phenotype. This will occur even if the cross involves a homozygous recessive plant, which has white flowers (pp) and a wrinkled pea shape (rr). However, if the pea plant is heterozygous for both traits (PpRr), then there is a 25% chance that the F_1 generation will show the recessive condition for one or both of the traits. Figure 16.14 shows a Punnett square involving a cross between heterozygous and homozygous recessive individuals. The expected ratio of individuals is one purple flower with round peas, one purple flower with wrinkled peas, one white flower with round peas, and one white flower with wrinkles peas (that is, 1 : 1 : 1 : 1).

What if you did a test cross by breeding a plant that was heterozygous for flower colour (Pp) and

DESIGN YOUR OWN
Investigation

16 • A

SKILL FOCUS

- Predicting
- Hypothesizing
- Identifying variables
- Performing and recording

Determining Plant Genotypes

In this investigation, you will design and conduct experiments to determine the genotypes of tobacco plant seeds. You will use two batches of seeds that yield slightly different characteristics in the plants. One batch of seeds will produce some seedlings that are green, and some that are white (albino). The other batch will produce some seedlings that are green, some green-yellow, and some yellow. Work in a small group to design your experiments. Then obtain your teacher's approval before completing the investigation.

Problem

How can you determine the genotype of a tobacco seed based on the phenotype of the seedling?

Hypothesis

Each group is responsible for formulating a testable hypothesis of how the plant phenotypes reflect their genotypes. The hypothesis will form the basis of your experimental design.

NOTE: Be careful not to mix seeds from the two batches. Wash your hands following this investigation.

Materials

2 different batches of tobacco seeds
flats, small pots, or plastic cups
growth medium (vermiculite or sterilized potting soil)
magnifying lens or dissecting light microscope
water
labels

Experimental Plan

1. Brainstorm several methods you could use to test your hypothesis, using the materials listed here.
2. As a group, select one method for your experimental design.
3. Your experimental design should include the collection of qualitative and quantitative data.
4. Your plan should consist of a series of easily identifiable and understandable steps that could be duplicated by another group, without the need for additional information or clarification.

Checking the Plan

Review your plan among the members of your group and with your teacher. Possible questions include:

1. What types of data will you collect?
2. What is/are the dependent and independent variable(s)? Does your experiment include any control variables? What variables might you wish to control?

homozygous recessive for pea shape (rr) with one that was homozygous recessive for both traits? In this case, all the offspring will show the recessive wrinkled pea shape. Each of the offspring has a 50% chance of inheriting the dominant allele for flower colour from the heterozygous parent. This means that half of the offspring will also be heterozygous for flower colour, while half will be homozygous recessive for flower colour. The expected ratio of offspring is therefore one purple flower with wrinkled peas to one white flower with wrinkled peas, or 1 : 1. The same ratio will result from a test cross involving a plant that is homozygous dominant for pea shape (RR) and heterozygous for flower colour (Pp). All offspring will have round peas, while approximately half will show the dominant flower colour.

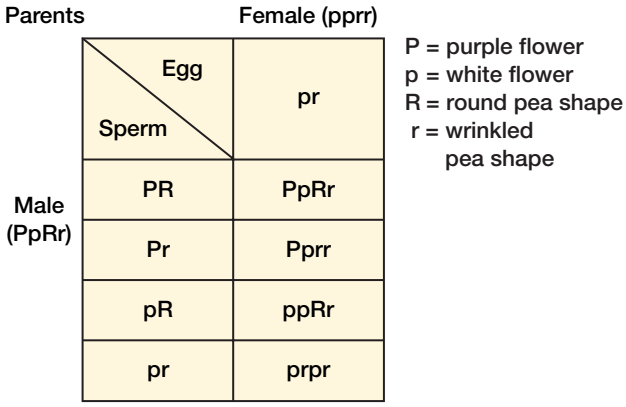


Figure 16.14 This two-trait test cross shows a cross between a heterozygous individual and a homozygous recessive individual. The Punnett square shows that there is a 25% chance of producing each of the possible phenotypes.

3. What is the duration of the experiment and what data will you collect during this time?
4. Have you prepared a table for collecting your data?
5. Has your plan been approved by your teacher?
6. Have you applied/used all necessary safety precautions?

Data and Observations

Each group is responsible for carrying out their own data collection. Record your observations in your table. Each group is also responsible for analyzing their results. Use a graph or chart to present your results.

Analyze

1. Suggest possible genotypes (combinations of alleles) for the different phenotypes observed (for example, GG might indicate homozygous dominant for green).
2. Why is it not possible for the genotypes of the different batches of tobacco plants to be determined through an investigation of the seeds alone?
3. Identify each of the variables you considered in designing your experiment. Explain how consideration of each variable was necessary in order to obtain valid scientific results.
4. Determine the method of inheritance for the batch of seeds that produced green and/or white tobacco seedlings. Explain.
5. Describe, how you determined the genotypes of the individual seeds using an example from each of the seed batches.

Conclude and Apply

6. Determine the method of inheritance for the batch of seeds that produce green, yellow-green, and/or yellow tobacco plants. How is this method different from the method you described above? Does this method of inheritance follow the principles laid out by Mendel? Explain.

Exploring Further

7. In this investigation, you looked at variations of a single trait — plant colour. How would you modify your experimental design to determine the genotypes of the seeds for two different traits?

Sample Problem

The Two-trait Cross

A male and a female guinea pig are both heterozygous for fur colour and fur texture. Both dark fur (D) and rough fur (R) are dominant traits.

(a) What are the recessive traits and what letters do you use for them?

(b) What are the parent phenotypes?

(c) How many different gametes are formed and what are they?

(d) Determine the frequency of offspring that are homozygous for both traits.

(e) Determine the frequency of offspring that have rough, dark fur.

(f) Determine the frequency of offspring that express both recessive traits.

What is required?

You are asked to determine the recessive traits and assign letters to represent them. Further, you are asked to determine the phenotypes of the parents and offspring.

What is given?

You know that both dark fur (D) and rough fur (R) are dominant traits. Therefore, the recessive traits are light fur (d) and soft fur (r). You are told that both parents are heterozygous. This means the genotype of each parent is DdRr. Because of the rule of dominance, each parent would have dark, rough fur.

Plan your strategy

To determine the genotype and phenotype of the offspring, you will need to make the following cross: DdRr × DdRr. A gamete from each parent can include one allele from each trait. Therefore, each parent can produce four possible gametes: DR, Dr, dR, and dr.

Act on your strategy

Place the gametes of each parent along the columns and rows of the Punnett square and complete the possible crosses.

Male gametes \ Female gametes	DR	Dr	dR	dr
DR	DDRR	DDRr	DdRR	DdRr
Dr	DDRr	DDrr	DdRr	Ddrr
dR	DdRR	DdRr	ddRR	ddRr
dr	DdRr	Ddrr	ddRr	ddrr

There are only two individuals homozygous for both fur colour and texture. These are DDRR and ddrr. Therefore, the proportion of offspring purebred for these traits is $\frac{2}{16}$ or $\frac{1}{8}$.

The following offspring have rough, dark fur: DDRR, DDRr, DdRR, and DdRr. There are nine individuals with this genotype. Therefore $\frac{9}{16}$ of the offspring have rough, dark fur.

Only one offspring is homozygous recessive (ddrr). Thus $\frac{1}{16}$ of all the offspring will have light, smooth fur.

Check your solution

The genotype of each parent is DdRr. This means that each parent will have dark, rough fur. A cross produces nine possible genotypes in the offspring: DDRR, DDRr, DdRR, DDrr, DdRr, Ddrr, ddRR, ddRr, and ddrr. The phenotypic ratio of the offspring is:

- $\frac{9}{16}$ dark and rough fur
- $\frac{3}{16}$ dark and smooth fur
- $\frac{3}{16}$ light and rough fur
- $\frac{1}{16}$ light and smooth fur

Practice Problem

1. In people, curly hair is dominant over straight hair and the ability to curl the tongue is dominant over not being able to curl the tongue. A man with curly hair who has the ability to curl his tongue and a woman with curly hair who cannot curl her tongue have children. What are the possible genotypic and phenotypic ratios of their offspring?

Beyond Mendel's Laws

In his studies of pea plants, Mendel found that inherited traits were either dominant or recessive. The dominant allele in an individual was always expressed, even if the recessive allele was present. However, some organisms show different patterns of inheritance. How can we explain the inheritance of traits that do not follow simple Mendelian genetics?

Incomplete Dominance

Not all traits are purely dominant or purely recessive. In some instances neither of the alleles controlling the trait are dominant. When this happens, a blending of the two traits can occur, called **incomplete dominance**. Examples of incomplete dominance can be found in many species of plants. For example, white or red snapdragon flowers are homozygous, while pink flowers are heterozygous. In this example, the letters R and R′ are used (rather than R and r) to indicate alleles that show incomplete dominance (see Figure 16.15). Two red alleles (RR) are necessary to produce a red flower.

Individuals with only one R allele are unable to make enough red pigment to produce red flowers, and they appear pink. Individuals that are white (R′R′) produce no red pigment.

Inheritance of traits that show incomplete dominance follows Mendel's laws of segregation and independent assortment. Because neither of the alleles is dominant, however, the offspring do not show a Mendelian ratio of phenotypes. Figure 16.15 shows the ratios of genotypes and phenotypes in F_1 and F_2 generations of snapdragon crosses.

Co-dominance

In some cases, both alleles for a trait may be dominant. Such alleles are said to be **co-dominant** because *both* alleles are expressed in the heterozygous individual. For example, feather colour in chickens is governed by two dominant alleles. Black birds are homozygous for the B allele and white birds are homozygous for the W allele.

What happens when a black rooster is crossed with a white hen? If the colours blended, you would expect offspring with grey plumage. If only the B allele was dominant, then only black-feathered young would result. However, the result of the cross is offspring with checkered black-and-white plumage, as shown in Figure 16.16. Some of the feathers are white and some are black.

Figure 16.16 In some varieties of chicken, two alleles for a trait may be expressed equally, such as in this bird with barred plumage.

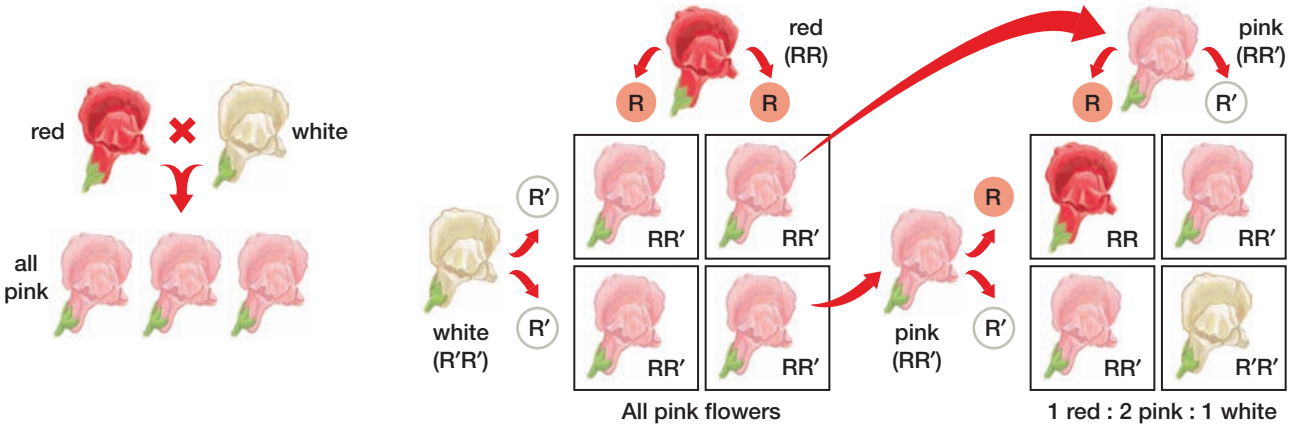


Figure 16.15 Flower colour in the snapdragon is an example of incomplete dominance. Pink flowers are heterozygous (*RR′*), where neither allele is dominant.

Multiple Alleles

Many genes have more than two alleles, or **multiple alleles**. An example of multiple alleles occurs in human blood types. In this case, three alleles are involved: A, B, and O. Table 16.1 shows the blood types in humans and the possible genotypes for each. Each person has two of the three alleles. The three alleles for human blood types are I^A, I^B, and i. Alleles I^A and I^B are dominant over i. However, I^A and I^B are co-dominant and are expressed equally.

There are two possible genotypes for blood types A and B, one homozygous, and one heterozygous. In order to have type AB blood, a person would have to inherit one I^A and one I^B allele. People with type O blood must inherit two ii alleles. You will learn about the inheritance of human blood-related illnesses in the next section.

Table 16.1
Human blood types

Phenotype (blood type)	Genotypes
A	I^AI^A or I^Ai
B	I^BI^B or I^Bi
AB	I^AI^B
O	ii

Sample Problem

Human Blood Types

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

What is required?

You are asked to determine the possible genotypes of offspring from a cross.

What is given?

You know the phenotype of the mother (AB) and father (A). The mother's genotype is therefore I^AI^B. The possible genotypes of the father are I^AI^A or I^Ai.

Plan your strategy

You must make the following crosses:
$I^AI^B \times I^AI^A$
$I^AI^B \times I^Ai$

Act on your strategy

Place the alleles of each parent along the columns and rows of the Punnett square and complete the possible crosses. If the father is blood type A homozygous, then:

Father \ Mother	I^A	I^B
I^A	I^AI^A	I^AI^B
I^A	I^AI^A	I^AI^B

If father is blood type A heterozygous, then:

Father \ Mother	I^A	I^B
I^A	I^AI^A	I^AI^B
i	I^Ai	I^Bi

There are four possible genotypes: I^AI^A, I^AI^B, I^Ai, I^Bi. (Note: the genotypes I^AI^A and I^Ai will both produce the same phenotype — type A blood). Thus, the possible blood types of the children are: A, AB, and B.

Check your solution

If the father is homozygous, the possible blood types of the children are A and AB. If the father is heterozygous, the possible blood types of the children are A, AB, and B.

Practice Problem

1. Suppose a man with blood type B marries a woman with type AB blood. What blood types would you expect to find among their children? What could tell you whether the man was homozygous or heterozygous for the B blood type?

Human blood type is only one example of a trait that is governed by more than one pair of alleles. Some traits are controlled by far greater numbers of alleles. These types of traits are the result of multiple gene inheritance and are very complex. Skin colour is a good example of this type of inheritance. Instead of just a few distinct phenotypes there is a continuous variation that cannot be split up into easily defined categories. The more genes that contribute to a single trait, the greater the number of categories of the trait, with increasingly fine differences between the categories.

Test crosses help to determine whether a particular trait is passed by one, two, or more pairs of alleles. Table 16.2 shows the phenotypic ratios of F_2 offspring produced after mating parents with contrasting traits and then breeding the F_1 individuals. Each ratio gives rise to a particular conclusion about the underlying genetic interactions. Why is it important to know how many pairs of alleles govern a particular trait? Multiple allele inheritance is often influenced by environmental factors, such as diet and climate, while single allele inheritance is not. Plant researchers, for example, need to know how much of their plant breeding results are due to genes and how much to the care they give growing plants. In the same way, medical practitioners treating genetic disorders need to know whether changes in diet or other behavior can help relieve the symptoms they find in their patients.

Table 16.2
Results of several test crosses

Phenotypic ratio in F_2 offspring	Conclusion
3 : 1	One pair of alleles at one gene location
9 : 3 : 3 : 1	Two pairs of alleles at two gene locations
No recognizable ratio	Alleles at multiple gene locations

THINKING LAB

Inheritance of Coat Colour in Rabbits

Background

Coat colour in rabbits is governed by four different alleles. Each allele is responsible for producing a different coat colour: dark grey, Chinchilla, Himalayan, and white. Each rabbit has only two alleles. Study the relationship among the alleles in the table and then complete the lab.

Phenotype (coat colour)	Allele	Pattern of inheritance
Dark grey	C	dominant to all other alleles
Chinchilla	c^{ch}	dominant to Himalayan and to white
Himalayan	c^{h}	dominant to white
White	c	recessive

You Try It

1. List all the possible genotypes for a
   - **(a)** dark grey rabbit
   - **(b)** Chinchilla rabbit
   - **(c)** Himalayan rabbit
   - **(d)** white rabbit
2. Predict the phenotype of a rabbit with the following genotypes. Explain your answers.
   - **(a)** $c^{h}c^{ch}$
   - **(b)** Cc^{h}
3. Would it be possible to obtain white rabbits if one parent is white and the other is Chinchilla? Explain.
4. Would it be possible to obtain Chinchilla rabbits if one parent is Himalayan and the other is white? Explain.
5. A Chinchilla rabbit is mated with a Himalayan. Some of the offspring are white. What are the parents' genotypes?

Pedigrees

Crosses can be done easily with plants, such as the common garden pea, in order to determine the inheritance of traits. How is inheritance of traits studied among humans? Because experimental crosses are not possible, human geneticists use medical, historical, and family records to study crosses that have already occurred. Records extending across several generations can be arranged in the form of a family **pedigree**. This is a diagram that illustrates the genetic relationships among a group of related individuals (see Figure 16.17).

In constructing a pedigree, squares are generally used to represent males and circles represent females. Shading or colour represents individuals who are recessive or dominant for a single trait (see Figure 16.17). Generations are indicated by roman numerals to the left of the pedigree. Some individuals may show only half a square or circle of colour. This indicates that the given individual is a **carrier**, heterozygous for the given trait but not showing it.

Since Mendel's time, knowledge of the mechanisms that control the inheritance of traits has developed considerably. In section 16.4 you will see how Mendel's laws and new knowledge about heredity are shaping the study of human genetics.

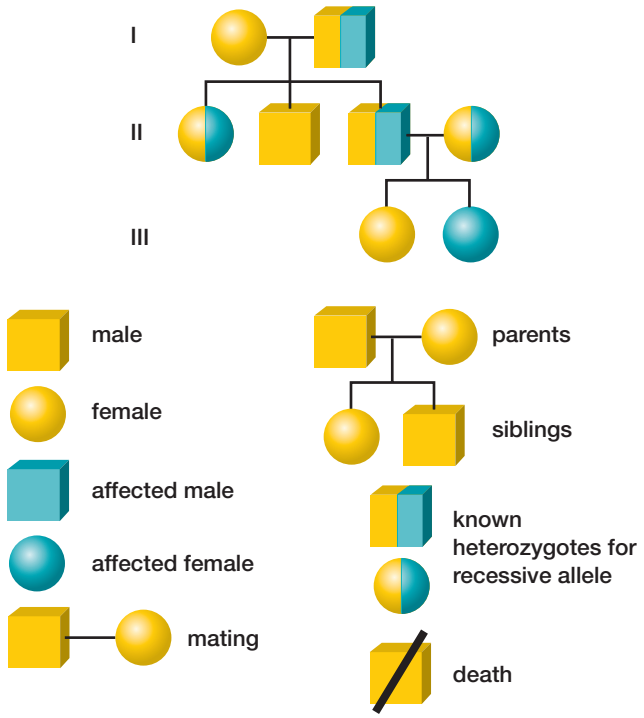


Figure 16.17 This example of a pedigree shows the method of transmission of dominant and recessive alleles from one generation to the next.

SECTION REVIEW

1. A dihybrid cross is made of pea plants. Purple flowers (P) are dominant and white flowers are recessive. Tall plants (T) are dominant and short plants recessive. Both parents are heterozygous for both traits. Prepare a Punnett square to determine the phenotype ratios of the offspring. What Mendelian law does this ratio demonstrate?
2. A rose-combed rooster is mated with two rose-combed hens. Hen A produces 14 chicks, all rose-combed. Hen B produces 9 chicks, 7 of which are rose-combed and 2 single-combed. What are the likely genotypes of the parent birds? Explain.
3. Define the following terms: carrier, incomplete dominance, co-dominance, multiple-allele inheritance.
4. How many different genotypes of the human blood type are possible? How many different phenotypes are possible? Explain why these numbers are different.
5. A woman sues a man for the support of her child. She has blood type A, her child has type O, and the man has type B. Could the man be the father? Explain.
6. In four o'clock plants, red flowers are incompletely dominant over white flowers. The heterozygous flowers are pink. If a red-flowered four o'clock plant is crossed with a white-flowered four o'clock plant, what will be the flower colour of
   (a) the F_1 generation?
   (b) the F_1 generation crossed with its white parent?
   (c) the F_1 generation crossed with its red parent?
7. A female hog may give birth to as many as 16 offspring in a single litter. If black coat colour in hogs is dominant to white, show how a breeder might expect an entire litter of piglets to have black coat colour.
8. Explain how a person can be a carrier for a particular disease and yet not have the disease. Is this true for all people who are carriers?

16.3 Chromosomes and Heredity

OUTCOMES

- **Describe the contributions of genetic scientists that led to the chromosome theory of inheritance.**
- **Solve sex-linked inheritance problems using Punnett squares.**
- **Demonstrate an understanding of gene expression and polygenic inheritance.**
- **Distinguish among different types of chromosome mutation.**

When Gregor Mendel formulated his laws of inheritance of traits, he did not know about meiosis or the existence of chromosomes in cells. Based upon his experiments with pea plants, Mendel concluded that each trait has two factors. During gamete formation, the factors segregate so that each gamete has one factor. Mendel's conclusions were remarkable for his time. However, his findings went largely unnoticed until the early 1900s, when better techniques were developed to investigate cellular processes and chromosomes were actually observed in cells.

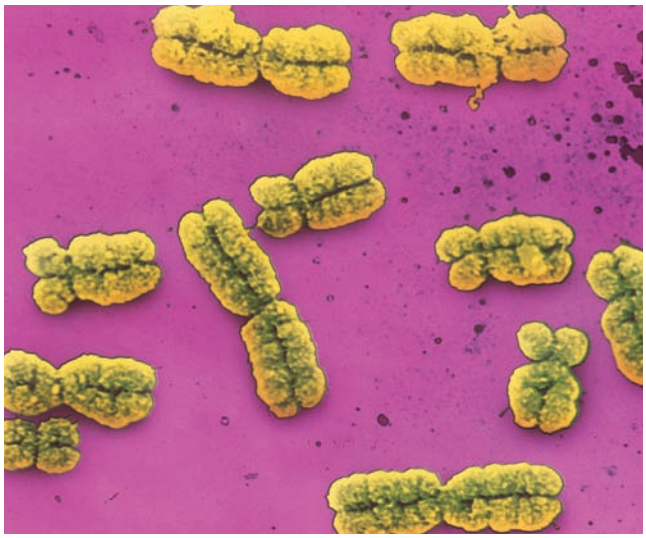


Figure 16.18 How are pairs of chromosomes related to pairs of alleles?

The Chromosome Theory of Inheritance

In 1902, scientists Walter Sutton and Theodor Boveri made an important discovery while studying the phases of meiosis. They realized that the behaviour of chromosomes during meiosis was related to the behaviour of "factors" in Mendel's experiments with pea plants. Figure 16.19 compares the behaviour of Mendel's factors and chromosomes.

Sutton and Boveri made three key observations. They noted that chromosomes occur in pairs, and that these pairs segregate during the anaphase I stage of meiosis. They also found that chromosomes align independently of one another along the equator of the cell. The result is that each gamete receives one chromosome from each pair, and this one chromosome does not influence which other chromosomes will be found in that gamete. These observations formed the basis for the **chromosome theory** of inheritance. The chromosome theory states that

- Mendel's factors, or genes, are carried on chromosomes.
- It is the segregation and independent assortment of chromosomes during meiosis that accounts for the patterns of inheritance.

As you will see later in this section, the chromosome theory can also account for patterns of inheritance that do not follow Mendel's laws.

Morgan's Discoveries

While investigating eye colour in fruit flies in 1910, American scientist Thomas Morgan produced a white-eyed male fly by crossing two red-eyed parent flies. This result was consistent with the law of dominance in a monohybrid cross. However, when Morgan crossed a red-eyed female offspring of the white-eyed male with a normal red-eyed male, all the female offspring of that cross had red eyes, while of the male offspring, half had red eyes and half had white eyes. The discovery that eye colour could be linked to sex led Morgan to deduce that the gene for eye colour was located on the X chromosome. This was the first time that a gene was linked to a specific chromosome.

Morgan also helped to explain why recombination occurs among some genes more frequently than others. In some testcrosses, Morgan found that certain genes did not follow the law of independent

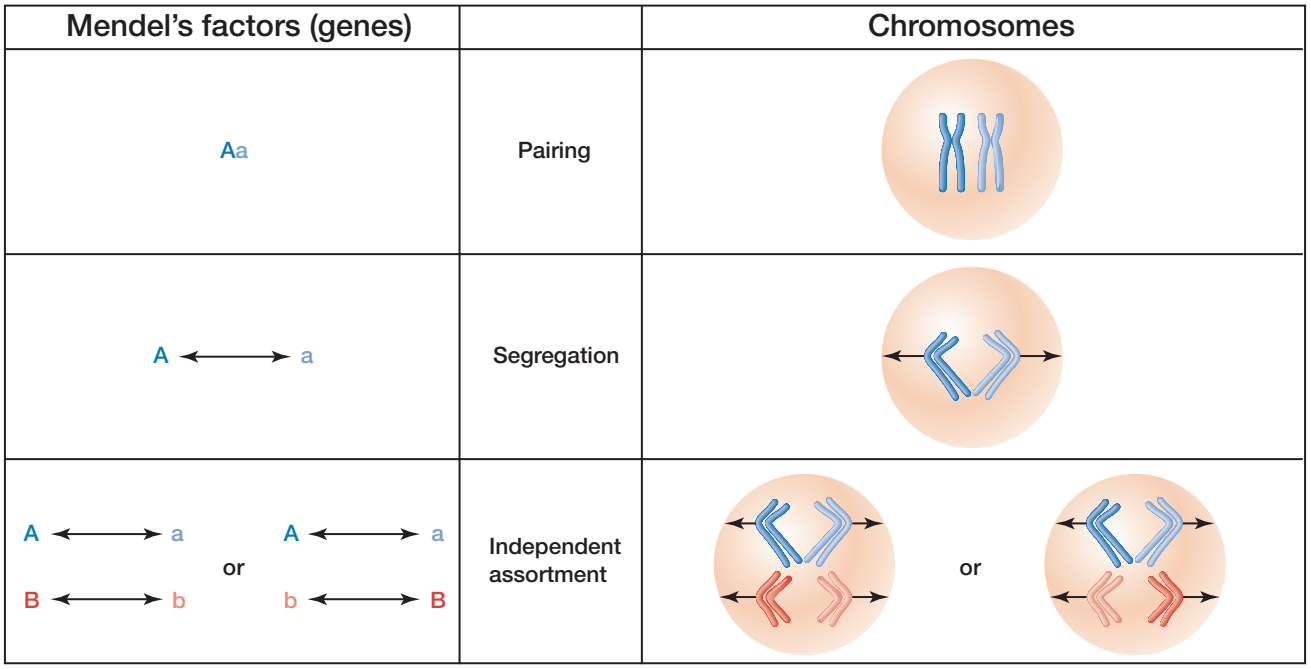


Figure 16.19 The behaviour of Mendel's factors (genes) and chromosomes during meiosis. Light and dark shades of the same colour show different members of the same homologous pair of chromosomes.

assortment, but instead tended to be inherited together. He reasoned that genes that are located on the same chromosome would usually not segregate. Such genes are called linked.

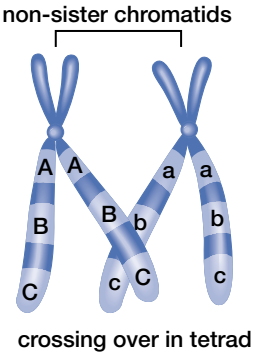


Figure 16.20 Morgan's work helped to explain why some linked genes recombine more often than others. Because of the greater number of potential crossover sites, crossing over is more likely to take place between the genes A and C than between genes A and B.

This rule does not always apply, however. In his studies, Morgan found that linked genes do sometimes segregate, and that the likelihood of the segregation varies among different pairs of linked genes. How is this possible? The **gene-chromosome theory**, which states that genes exist at specific sites arranged in a linear fashion along chromosomes, accounts for this observation.

Recall from Chapter 14, section 14.2 that crossing over can occur among chromosomes during cell division. As shown in Figure 16.20, genes located very close together along a chromosome will almost always be inherited together, while genes located some distance apart are more likely to be separated by a crossing-over event. The statistical likelihood of crossing over increases with the distance between the two genes. Morgan's work indicates that Mendel's law of independent assortment can be restated as *if crossing over does not take place, genes that are located on different chromosomes will assort independently while genes that are located on the same chromosome will be inherited together.*

Sex-Linked Inheritance

Some traits that are passed from one generation to the next depend on the sex of the parent carrying the trait. This is because the genes for these traits are located on the sex chromosomes. The transmission of genes that are located on one of the sex chromosomes, X or Y, is called **sex-linked inheritance**. A gene that is located on the X chromosome only is called X-linked. A gene that is located on the Y chromosome only is called Y-linked. Most of the known sex-linked traits are X-linked. Very few Y-linked traits are known. This may be because the Y chromosome is much smaller than the X chromosome.

Morgan's work provided the first explanation of the inheritance pattern for sex-linked traits. He hypothesized that the gene coding for eye colour in fruit flies was located on the X chromosome, as shown in Figure 16.21. He reasoned that the white-eyed trait was recessive. Therefore, the only way to obtain a female white-eyed fruit fly was if both the male and female parent donated an allele that coded for white-eyes. Figure 16.22 shows a cross between a white-eyed male (genotype X^rY) and a red-eyed female (genotype X^RX^R). The male donates the Y chromosome (which does not carry the eye colour gene) to all male offspring. The female, therefore, donates the X chromosome to the male offspring. The female offspring all receive the male parents X chromosome and one of the X chromosomes from the female parent. Therefore, all of the female offspring are heterozygous and have red eyes (X^RX^r).

BIO FACT

Many scientists believe that the Y chromosome originated when a mutation caused one arm of the X chromosome to break off close to the centromere.

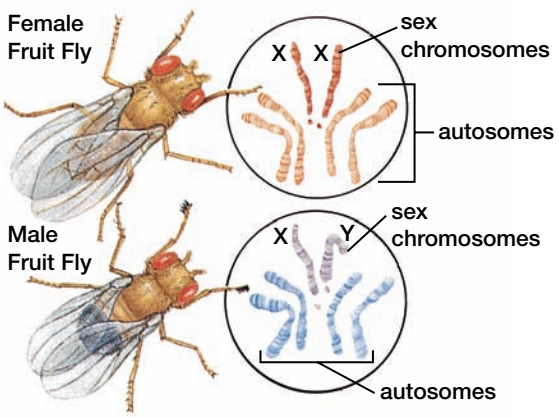


Figure 16.21 In fruit flies (*Drosophila melanogaster*), the gene for eye colour is located on the X chromosome.

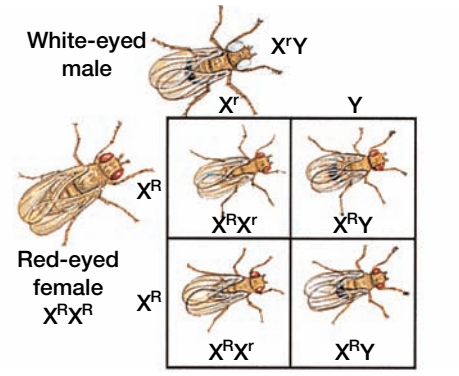
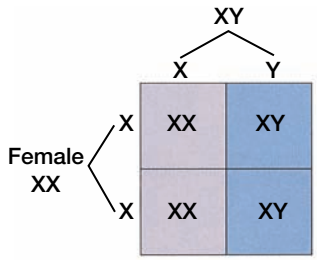


Figure 16.22 Crosses of fruit flies for red eyes (R) and white eyes (r).

Sex-linked Traits and Punnett Squares

Punnett squares can be used to predict the outcome of crosses involving genes that are located on the X or Y chromosome. When creating a Punnett square to determine the outcome of a cross involving a sex-linked trait, assume that the trait is located on the X chromosome unless it is stated that the trait is located on the Y chromosome. As shown in Figure 16.23, the gametes containing the X and Y chromosomes are placed on the outside of the Punnett square in the same manner as autosomal traits. The only differences are that the alleles are written on the X chromosome if the trait is X-linked and no allele is written with the Y chromosome. When the genotype and phenotype ratios are written, the sex is always included in the ratio.



Genotypic ratio = 1/2 XX : 1/2 XY
Phentypic ratio = 1/2 female : 1/2 male

Figure 16.23 In this Punnett square, half of the offspring are female and half are male.

Chromosomes and Gene Expression

As you have seen, many patterns of inheritance do not follow Mendelian ratios. In some cases, for example, genes do not follow the law of independent assortment. This can happen when the genes being studied are located on the same chromosome, which means that they will be inherited together unless they are separated by crossing over during meiosis (for a review of crossing over, see Chapter 14). The following pages describe some other inheritance patterns that can be explained by the chromosome theory of inheritance.

Chromosome Inactivation

Males and females produce the same amounts of proteins coded by genes located on the X chromosome. However, females have two copies of the X chromosome in every cell and males only have one copy of the X chromosome in every cell. Experimental evidence has demonstrated that one of the X chromosomes in each female cell is inactivated. Which one is inactivated is random, and therefore different X chromosomes are active

in different cells. The inactivated X chromosome is called a **Barr body**.

The tortoiseshell coat colour in cats is an example of the presence of inactivated X chromosomes. The tortoiseshell pattern can only occur in female cats (see Figure 16.24). Each tortoiseshell cat has a random distribution of orange and black patches. The allele for orange or black coat colour is located on the X chromosome. A male cat will have only one allele for coat colour, either X^OY or X^BY, and so will have either orange or black fur (often mixed with white). A female cat, however, can carry alleles for both orange and black fur: X^OX^B. The pattern of Barr bodies in the hair cells of the cat results in a patchwork of orange and black fur. That is, the orange patches are made up of cells in which the X chromosome carrying the allele for black fur has been deactivated, and vice versa.

Figure 16.24 Only female cats show the tortoiseshell fur colour pattern. Why is this so?

DESIGN YOUR OWN
Investigation

16 • B

SKILL FOCUS

- Initiating and planning
- Hypothesizing
- Identifying variables
- Performing and recording
- Analyzing and interpreting

Measuring Variation in a Trait

Different traits show different degrees of variation. Some traits are either present or absent, while others show continuous variation. Traits that are either present or absent may be governed by a single gene, whereas traits that show continuous variation may be governed by several or many genes. The graphical representation of a trait can provide information about its inheritance pattern. How can the inheritance pattern of a trait be graphically represented? In this investigation, you will design a method to determine the inheritance pattern of two human traits based on data you will collect.

Problem

How can you infer the inheritance pattern of a trait?

Hypothesis

Make a hypothesis about the inheritance pattern of a trait based on the shape of the trait's graph. The graph should illustrate the variation of the trait on the *x*-axis and the number of students on the *y*-axis.

NOTE: Follow your teacher's directions for conducting this investigation in a manner that is always respectful of others.

Materials

Ruler or measuring tape

Experimental Plan

1. Prepare a list of the possible ways you might test your hypothesis.
2. Decide on one approach for your investigation that could be conducted in the classroom.
3. Your design should test one variable at a time. Plan to collect quantitative data, and prepare a table that will organize the data effectively.
4. Outline a procedure for your experiment listing each step. Provide a list of your materials and the quantities you will require. Get your teacher's approval before conducting your investigation.

Checking the Plan

1. What will you measure or count? What kind of graph will you produce using the data?

Polygenic Inheritance

Many traits are controlled by more than one gene. This phenomenon is known as **polygenic inheritance**. The result of many genes working together is a range of variation, or **continuous variation**, in a trait. Continuous variation can be defined as the variation among individuals in a population in which there is a gradient of phenotypes for one trait. An example of continuous variation is the range of ear length in corn. Figure 16.25 shows that the longest ear length is obtained when both genes controlling corn ear length are homozygous dominant for both alleles. The shortest ear length occurs when both genes controlling corn ear length are homozygous recessive for both alleles. As indicated by the Punnett square, the longest and shortest phenotypes are the least common because each can only be obtained by one combination of alleles. The medium length is most common because it can be obtained through many different combinations of alleles. In humans, height and skin colour are thought to be polygenic traits. In the following investigation, you will use graphical methods to infer the inheritance pattern of several human traits.

BIO FACT

Some genes are controlled through a process called imprinting. This occurs when a series of chemical markers produced by some genes are attached to other genes to make them become inactive.

Modifier Genes

Some genes, called **modifier genes**, work together with other genes to control the expression of a trait. The expression of eye colour in humans may involve modifier genes. For example, brown eye colour is the result of the presence of the pigment called melanin that is coded for by the dominant allele for eye colour. Blue eye colour is the result of the absence of the pigment melanin coded for by

2. How will you infer the inheritance pattern of a trait from the shape of a graph?

Data and Observations

Conduct your investigation and make your measurements. Enter the data into your table. Make a graph of your results.

Analyze

1. Compare the shape of the graphs for the two human traits you investigated.
2. Suggest reasons for the similarity or difference in the shapes of the two graphs.
3. Compare the shape of the your graphs to the graphs drawn by other students.
4. Suggest reasons for the similarities or differences in the shapes of the graphs.

Conclude and Apply

5. Based on your results, what inferences can be drawn about the inheritance pattern of a trait from the shape of the graph?

Exploring Further

6. What is a normal distribution? How do the results you obtained in this investigation compare with a normal distribution? Explain briefly.
7. The range of variation of the traits you investigated may be the result of many factors. Carry out research to investigate which factors contribute the most, and which factors contribute least, to the variation of the traits that you investigated. From your findings, what could an individual do to influence the development of the traits you investigated?

COMPUTER LINK

Use a spreadsheet program to input the data for the trait being measured. Generate the graphs using the spreadsheet program.

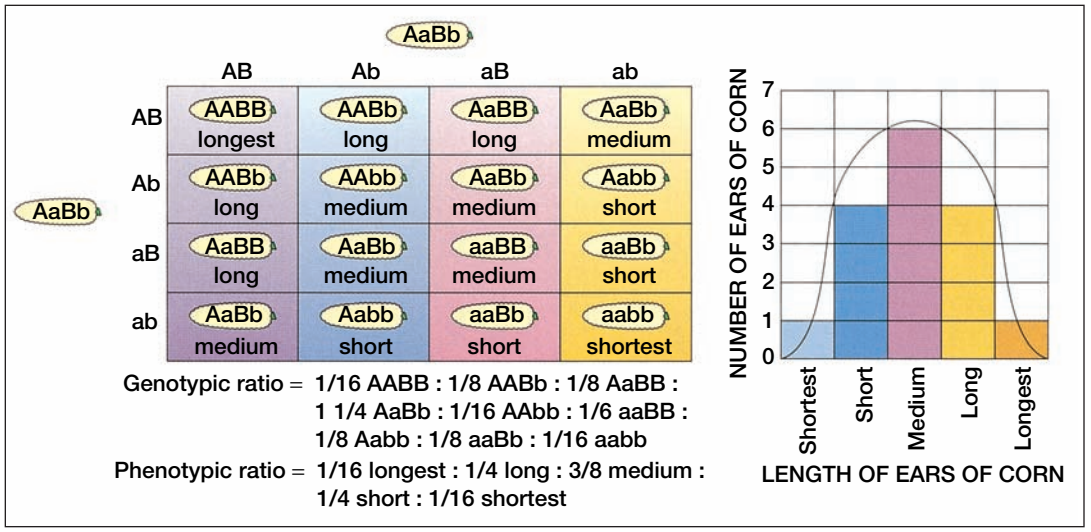


Figure 16.25 Ear length in corn is determined by two pairs of genes. The F_2 generation shows continuous variation in this trait, with medium length ears occurring most often.

the recessive allele for eye colour. Various modifier genes help to produce other eye colours.

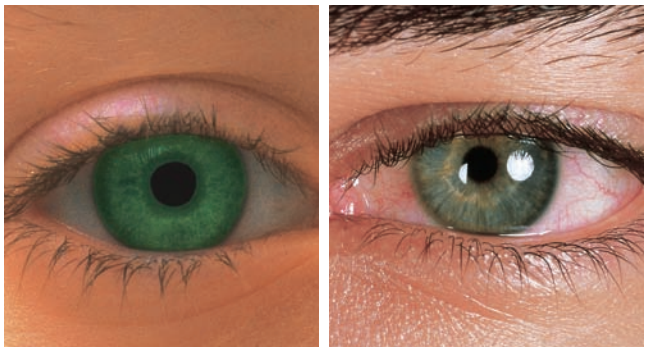


Figure 16.26 The gene for eye colour only codes for the presence or absence of the melanin pigment, which produces either brown or blue eyes. If this is the case, why are there other eye colours in humans?

Changes in Chromosomes

The patterns of inheritance you have studied so far reflect the nature of the genes that are associated with particular traits. Since these genes are located on chromosomes however, some genetic characteristics are associated with changes that occur at the level of entire chromosomes. The following pages describe how these changes can occur.

Changes in Chromosome Structure

Significant changes in the physical structure of chromosomes can occur either spontaneously or when cells are irradiated or exposed to certain chemicals. Essentially, there are four means by which these changes can happen: deletion, duplication, inversion, and translocation, as shown in Figure 16.27.

In a **deletion**, a portion of the chromosome is actually lost. There are several factors that can

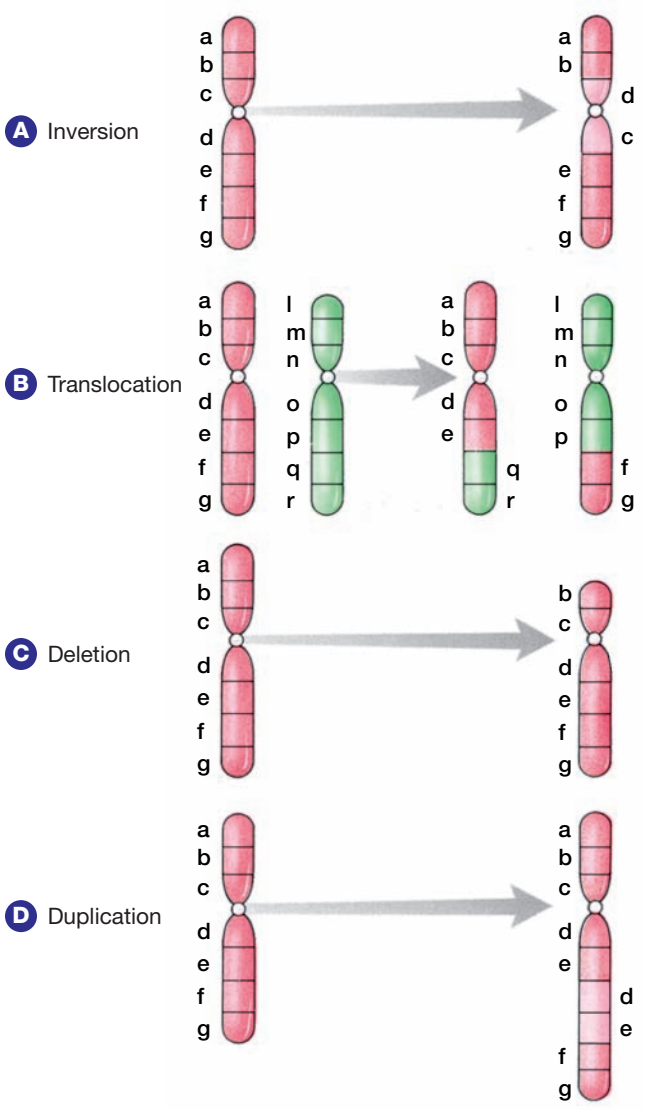


Figure 16.27 Means by which chromosome structures can change

trigger a deletion. Viruses, irradiation, and chemicals can cause pieces of a chromosome to be broken off. These pieces contain genes, and with them go needed genetic information. For example, when a piece of chromosome 5 is lost, a child is born mentally handicapped and with differences in facial appearance. Because an abnormally developed larynx makes the affected infant's cries sound like the mewing of a cat, this disorder is called *cri-du-chat* (French for cat's cry).

When an **inversion** occurs, a certain gene segment becomes free from its chromosome momentarily before being reinserted in the reverse order. This completely changes the position and order of the chromosome's genes, and can alter gene activity. Autism is a mental disorder originating in infancy. Some forms of autism are thought to be associated with chromosome inversions. Although they may be exceptionally bright and creative, autistic individuals have difficulty communicating or forming relationships with others. Symptoms of the disorder also include repetitive behavior such as rocking back and forth.

A **duplication** occurs when a gene sequence is repeated one or more times within one or several chromosomes. Even though certain gene sequences can be repeated thousands of times in normal chromosomes, there appears to be a point at which too many repeats affect the functioning of the gene. For example, in a condition called **fragile X syndrome** (which affects approximately one in 1500 males and one in 2500 females), a duplication occurs in chromosome X (see Figure 16.28). A specific sequence in a specific gene has been studied in affected persons. Most people have about 29 repeats of this specific sequence; people

Canadians in Biology

Pioneer Cytogeneticist

Random chance lies at the heart of genetics. Chance unions of gametes result in genetically unique children. Chance is also at the heart of the life and scientific career of Dr. Irene Ayako Uchida, a Canadian pioneer in cytogenetics, or the study of the chromosomes in cells.

Dr. Irene Uchida working in the laboratory.

Empathy and Science

Irene Uchida was born in Vancouver in 1917 as one of four daughters of Japanese immigrant parents. Her path into genetic science was quite unusual, especially for a woman of Japanese ancestry in the British Columbia of her day.

In 1942, with Canada and Japan at war, Uchida had almost finished her Bachelor of Arts degree at the University of British Columbia when she was suddenly evacuated from her Vancouver home to "Ghost Town" in the B.C. interior, near Lemon Creek. She acted as principal of the largest Japanese internment camp school in the Kootenays. This challenging experience nudged Uchida toward a career in social work when she resumed her studies at the University of Toronto after the war.

Shortly before she graduated, one of her professors suggested she pursue genetics. A Ph.D. in Zoology followed in 1951, after which Dr. Uchida began work at Toronto's Hospital for Sick Children as a Research Associate.

In 1959, Dr. Uchida moved to the University of Wisconsin and began laboratory studies of human chromosomes and their abnormalities. She wanted to know why Down syndrome children had an extra chromosome. She discovered that there appeared to be a link between maternal radiation received before conception and the extra chromosome 21 found in Down syndrome children. She then started to focus on chromosome 18 trisomy, which usually results in miscarriages.

Building Canada's Genetics Foundation

Today, Dr. Uchida's cytogenetic research is seen as an important forerunner of the ongoing Human Genome Project, to which her Canadian successors have made significant contributions. Her many honours include being named a Woman of the Century in 1967, one of 25 Outstanding Women by the Ontario Government during International Women's Year in 1975, and an Officer of the Order of Canada in 1993.

Dr. Uchida suggests that students approach a career in science with the sort of feistiness and joviality that served her so well. As genetics does, she believes in letting the creative power of random chance help shape the course of your life.

with fragile X syndrome have about 700 repeats of the same sequence.

Figure 16.28 The arrow indicates the fragile site on this fragile X chromosome.

In **translocation**, part of one chromosome changes places with another part of the same chromosome or with part of another, nonhomologous chromosome. If a part of chromosome 14 exchanges places with a part of chromosome 8, cancer can occur in the affected individual. Similarly, some occurrences of Down syndrome are related to translocation between chromosomes 14 and 21, while one kind of leukemia can be traced to translocation between chromosomes 22 and 9.

BIO FACT

Eighteen of our chromosome pairs are identical to those of gorillas and chimpanzees. The other five pairs differ due to inversions and translocations. These differences seem to be responsible for the differences among the primates.

Nondisjunction

Some genetic conditions and disorders are caused when chromosomes or chromatids do not separate as they should during meiosis. You will recall from Chapter 14 that this phenomenon is called nondisjunction. If nondisjunction occurs in meiosis, the gametes will have either too many or too few chromosomes. If one of these gametes is involved in fertilization, the result will be an embryo with extra or fewer chromosomes than normal. Inheriting an extra chromosome is called trisomy. Inheriting only one chromosome instead of a pair of chromosomes is called **monosomy**. Human embryos with too many or too few autosomes rarely survive. Embryos that were miscarried (spontaneously aborted) often have one extra or one fewer autosomes than normal.

WEB LINK

www.mcgrawhill.ca/links/atlbiology

A variety of human genetic disorders are the result of changes in chromosome structure. To find out more about the causes and effects of these changes, go to the web site above, and click on **Web Links**. What other organisms are commonly used to study changes in chromosome structure? What methods are used to detect changes in chromosome structure? Choose one type of chromosomal structural change. What effects does the chromosome change have on the phenotype of the individual? Can these symptoms be treated? Write an abstract (summary) highlighting the scientific findings in the research area you have chosen.

THINKING LAB

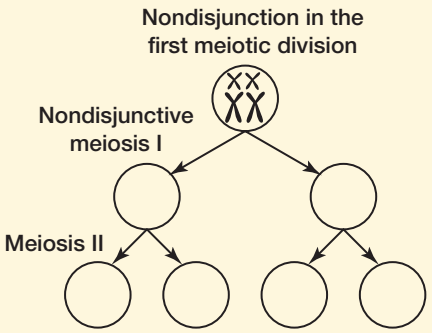
Modelling Nondisjunction

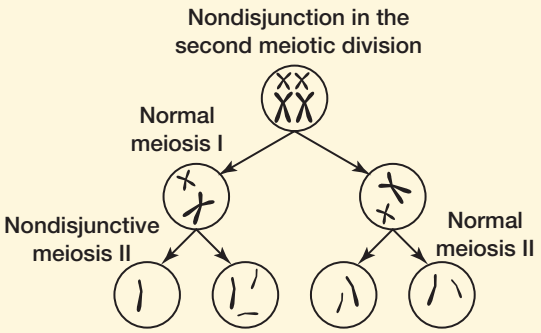
Background

Nondisjunction during meiosis can occur during either anaphase I or anaphase II. During these stages, the centromere splits and the spindle fibres shorten to pull the chromatids to opposite poles of the dividing cell. In this lab, you will apply what you have learned to explore the various types of gametes that can result from nondisjunction and how they relate to certain genetic disorders.

You Try it

1. The diagram on the right shows the gametes that would result if nondisjunction occured at meiosis II. Copy the diagram on the left into your notebook, and complete it to show what would happen if nondisjunction occured at meiosis I.





One of the most common genetic disorders resulting from nondisjunction during gamete formation is **Down syndrome**, or trisomy 21. Down syndrome arises when an individual receives three rather than two copies of chromosome 21. It is called a **syndrome** because it involves a group of disorders that occur together. Although symptoms can vary from one person to another, most individuals have mild to moderate mental impairment and a large, thick tongue that can create speech defects. In addition, the skeleton may not develop properly, resulting in a short, stocky body type with a thick neck.

BIO FACT

Other trisomies that occur in addition to Down syndrome include trisomy 8, trisomy 13 or Patau syndrome, trisomy 18 or Edward syndrome, and trisomy 22. Infants born with these trisomies have a host of serious physical conditions. Those born with trisomy 13 or 18 seldom live more than a few months.

Individuals with Down syndrome are more susceptible to infections and usually have abnormalities in one or more vital organs. About 40% have heart defects. Surgery early in life has proven to be effective in correcting tongue and facial defects. Individuals can also participate in many everyday activities. They may survive into their thirties or forties and beyond, but have a greater chance of developing a form of senility similar to Alzheimer's.

Nondisjunction can also occur among the sex chromosomes. When this happens, an individual receives too many or too few X or Y chromosomes. **Turner syndrome** results when a person inherits only a single X chromosome and no Y chromosome. An XO woman with Turner syndrome will be infertile. She will have female external genitalia but no ovaries and therefore no menstrual period, and will fail to mature sexually. She will also have other developmental defects, including a webbed neck, heart defects, kidney abnormalities, and skeletal abnormalities. Other problems can include learning difficulties and thyroid dysfunction. There is no comparable syndrome for the inheritance of only a single Y chromosome and no X chromosome, because YO embryos are not viable.

The inheritance of an extra X chromosome does not typically have significant effects on women, but an XXY male is likely to have Klinefelter syndrome. Individuals with this syndrome will have immature male sexual organs and will not grow facial hair. They are also likely to show some breast development. XYY males typically have Jacobs syndrome. Symptoms of Jacobs syndrome include speech and reading problems and persistent acne.

Nondisjunction can happen by chance in any cell, but some factors influence the probability of certain nondisjunctions. Maternal age, for example, is strongly linked to the incidence of Down syndrome. In the next section, you will see how patterns of inheritance in families provide information about the nature of genetic disorders.

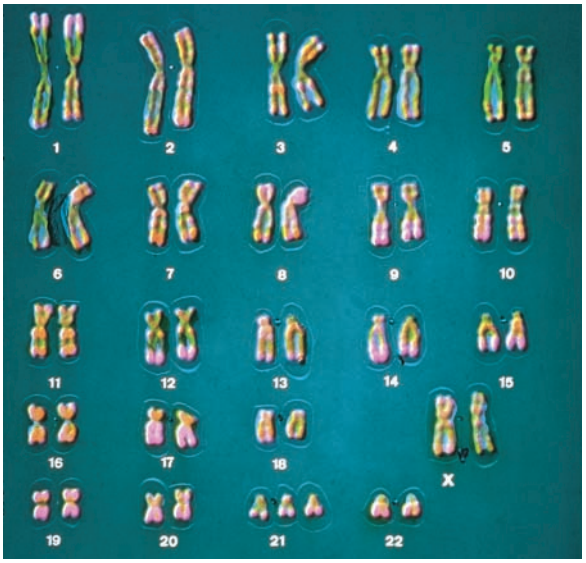


Figure 16.29 A child with Down syndrome at the Special Olympics. Can you spot the additional chromosome in the karyotype?

SECTION REVIEW

1. State the law of independent assortment. Explain briefly what is meant by independent assortment.

2. In a dihybrid cross of two pea plants, one homozygous for two dominant traits and the other homozygous for the corresponding recessive traits, what will the phenotypic ratio be for the F_1 generation? for the F_2 generation?

3. Give the possible alleles in the eggs produced by a woman whose genotype is JjKkLl.

4. In *Drosophila melanogaster* (the fruit fly you find hanging around your vegetables and fruits at home) normal wings (W) are dominant over vestigial wings (w); grey body colour (G) is dominant over ebony colour (g); and normal antennae (A) are dominant over antennapedia (a). Bugsy, a male fruit fly who is homozygous dominant for body colour and normal wing shape, mates with Daisy, a female fruit fly who is homozygous recessive for body colour and wing shape.

(a) Write the genotypes of Bugsy and Daisy for body colour and wing shape. Bugsy and Daisy will be the P generation.

(b) What are the alleles that Bugsy can provide for body colour? for wing shape?

(c) What are the alleles that Daisy can provide for body colour? for wing shape?

(d) Draw a Punnett square and show the possible genotype(s) of the F_1 generation produced by Bugsy and Daisy.

(e) What are the percentages of each genotype?

(f) What are the alleles for body colour and wing shape that can be provided by any member of the F_1 generation?

(g) Now cross two individuals from the F_1 generation (a dihybrid cross). Show this cross. The offspring from this cross will be the F_2 generation.

(h) What are the genotypic ratios for body colour in the F_1 generation? for wing shape?

5. Fruit with seeds are dominant over fruits that are seedless, and blue colour is dominant over purple colour. A homozygous purple fruit with seeds is crossbred with a homozygous blue, seedless fruit. What are the genotypes and phenotypes of the F_2 generation?

6. What observations did Sutton and Boveri make with respect to the behaviour of chromosomes? Why was this significant?

7. State the chromosome theory of inheritance and explain the relationship between Mendel's "factors" and chromosomes.

8. In fruit flies, red eye colour is an X-linked trait. Red eyes (R) are dominant over white eyes (r). Using a Punnett square, predict the genotypic and phenotypic ratios of the F_1 generation from

(a) a cross between a red-eyed male fruit fly and a white-eyed female fruit fly

(b) a cross between a red-eyed female fruit fly and a white-eyed male fruit fly

9. A normal chromosome contains the following sequence of genes: ABCDE. How might the sequence change if the following events occur?

(a) a deletion

(b) a duplication

(c) an inversion

(d) a translocation

10. Using diagrams, illustrate the chromosomal event that leads to Down syndrome.

11. Explain two different circumstances that could produce a range of phenotypes from a single trait.

16.4 Human Genetics

OUTCOMES

- **Distinguish among different types of autosomal and sex-linked inheritance patterns.**
- **Identify and describe the causes of several types of human genetic disorders.**
- **Analyze and interpret models of human karyotypes.**
- **Draw and interpret patterns of inheritance shown on human pedigree charts.**

The study of human genetics is complicated by a number of factors. Humans have long lifespans and produce very few offspring in comparison with plants or fruit flies. Furthermore, most people do not keep accurate records of their family history. Only in rare cases, such as that of the British royal family shown below, have detailed records been kept for over a century.

Family pedigrees show that some traits are inherited according to the principles that Mendel described. Some traits are sex-linked, while others are carried on autosomes, which are any of the remaining 22 pairs of chromosomes that make up the human genome. Other genetic traits arise with changes in chromosomes. The following pages describe some of the patterns of inheritance of human genetic disorders.

Figure 16.30 The British royal family is one of the few human families for which a detailed pedigree exists.

Autosomal Recessive Inheritance

There are many autosomal recessive disorders. Such disorders are carried on the autosomes and are not specific to the sex of the person. One example of such a disorder is **Tay-Sachs disease**. Children with Tay-Sachs disease appear normal at birth; however, their brains and spinal cords begin to deteriorate at about eight months of age. By their first birthday, these children are blind, mentally handicapped, and display little muscular activity. Most die before their fifth birthday.

Individuals with Tay-Sachs disease lack an enzyme in the lysosomes of their brain cells. Lysosomes are cell organelles in which large molecules are digested. The recessive allele does not code for the production of the enzyme responsible for breaking down specific lipids inside the lysosomes. As undigested lipids build up inside the affected person, the lysosomes become enlarged and eventually destroy the brain cells that house them (see Figure 16.31).

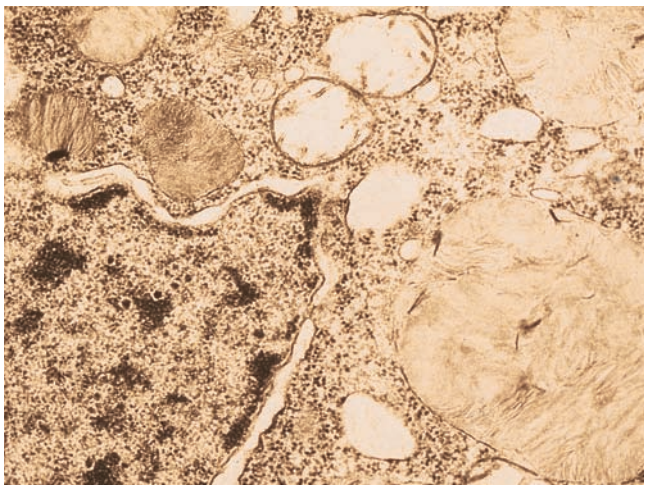


Figure 16.31 Electron micrograph of brain tissue of a person affected with Tay-Sachs disease shows enlarged lysosomes filled with lipid deposits. An enzyme deficiency prevents these deposits from being degraded.

There is no treatment for Tay-Sachs disease. However, a blood test has been developed to identify heterozygous carriers. Carriers have half the enzyme levels of normal individuals, which is enough to function normally.

MATH LINK

You can use simple Mendelian genetics to determine if a condition or disorder is due to an autosomal recessive allele. If both parents are heterozygous carriers of a recessive allele, what proportion of their children will be at risk of inheriting both copies of the allele? What proportion will be at risk if both parents are affected, meaning that they are both homozygous recessive? Construct Punnett squares and present your findings as genotype and phenotype ratios.

Another autosomal recessive disorder that affects young children is **phenylketonuria (PKU)**. In individuals with this condition, an enzyme that converts phenylalanine to tyrosine is either absent or defective. Phenylalanine is an amino acid essential for regular growth and development, and for protein metabolism. Tyrosine, another amino acid, is used by the body to make melanin and certain hormones. The phenylalanine in children with PKU is broken down abnormally, and the products of this process damage the developing nervous system.

Babies with phenylketonuria appear normal at birth. If their condition is not diagnosed and treated, however, they will become severely mentally handicapped within a few months. Fortunately, newborns today are routinely tested for PKU. Infants who test positive for the disorder are placed on a special diet that prevents the harmful products from accumulating. Once their nervous systems are fully developed, these individuals can go on to lead healthy lives.

Albinism is a genetic condition in which the eyes, skin, and hair have no pigment. The colour of our hair, skin, and eyes is due to varying amounts of a brown pigment called *melanin*, which is produced in special pigment cells. People who are homozygous for this autosomal recessive allele either lack one of the enzymes required to produce melanin or, if the enzyme is present, lack the means to get the enzyme to enter the pigment cells. Because skin and eye pigment plays an important role in protecting the skin and retina against harmful rays from the Sun, albino people and animals face a high risk of sunburns and eye damage from exposure to the Sun.

Figure 16.32 Albinism occurs among humans and many other organisms. People with this autosomal recessive condition have white or fair hair, and pale blue or pink eyes.

Codominant Inheritance

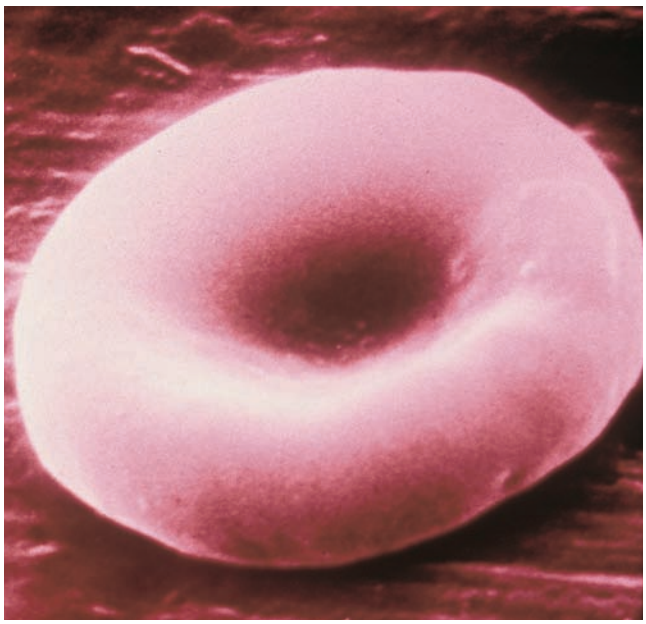
Sickle cell anemia is one of the better-known examples of a codominant genetic disorder. Affected individuals have a defect in the hemoglobin in red blood cells. This defect leads to blood clots and reduced blood flow to vital organs. As a result, they have little energy, suffer from various illnesses, and are in constant pain. Many die prematurely.

Heterozygous Advantage

The recessive allele that causes sickle cell anemia is thought to have originated in Africa. Until recently, homozygous recessive individuals never survived to become parents, indicating that the recessive allele was constantly being removed from the population. Yet in some African regions, almost half the population is heterozygous for sickle cell anemia. Geneticists wondered how this allele could remain at such high levels when it was constantly being removed from the population.

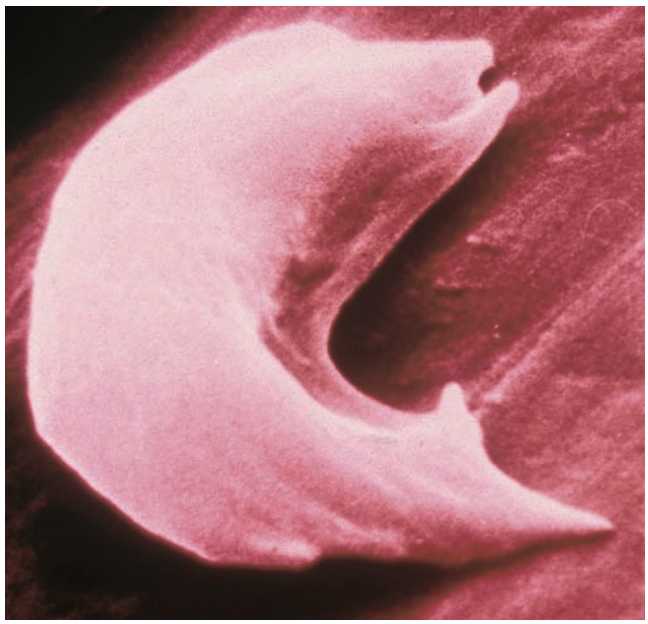
The answer came from studying another serious disease in the regions where sickle cell anemia is most commonly found. In Africa, malaria is a leading cause of illness and death, particularly among the young. Studies revealed that children who were heterozygous for sickle cell anemia were less likely to contract malaria and therefore more likely to survive to parenthood.

This is a classic example of **heterozygous advantage**, in which individuals with two different alleles for the same trait have a better rate of survival.



Magnification: 90 000 x

Red blood cells containing normal hemoglobin are round and smooth, allowing them to pass through capillaries easily.



Magnification: 90 000 x

Sickled red blood cells have elongated, blunt shapes that stick easily in capillaries and clog them.

Figure 16.33 Electron micrographs of normal and sickled red blood cells.

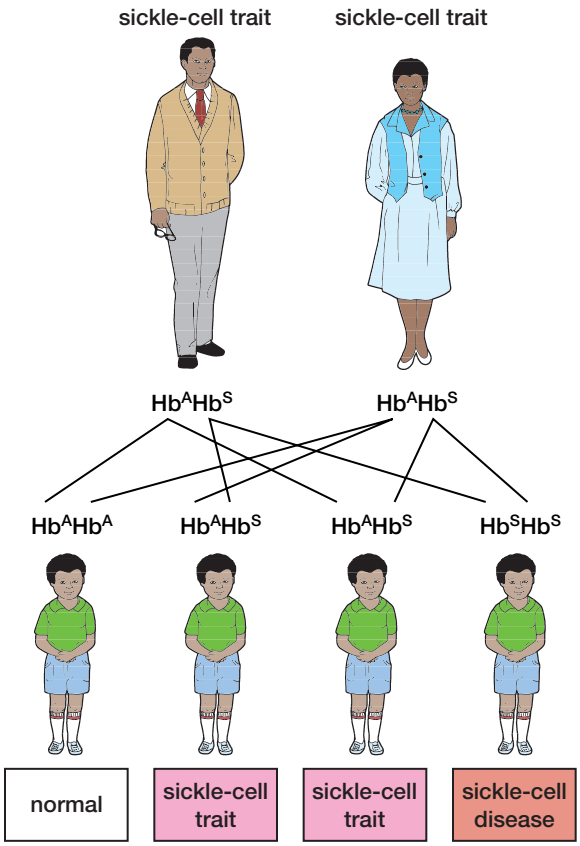


Figure 16.34 Inheritance of sickle cell anemia. In this example, each parent is heterozygous for the sickle cell trait. Among the offspring, there is a 50% chance of inheriting the sickle cell trait, a 25% chance of having sickle cell anemia, and a 25% chance of not having the disease.

Autosomal Dominant Inheritance

Researchers can use two pieces of evidence from Mendelian genetics to determine if an autosomal dominant allele is responsible for a trait. First, since a dominant allele is expressed in heterozygotes as well as in homozygous dominant individuals, the trait will appear in every generation. Second, if one parent is heterozygous and the other is homozygous recessive for the allele, then 50% of the offspring will have the trait.

Although genetic disorders caused by autosomal dominant alleles are very rare in human populations, they continue to exist. Some of them are caused by rare, chance mutations. In other cases, symptoms arise only after affected individuals have passed the age at which most of them have had children. The Punnett square in Figure 16.35 shows how an autosomal dominant trait can be inherited.

Progeria is a rare disorder that causes an individual to age rapidly. Progeria affects one in eight million newborns and does not run in families. This indicates that this very unusual affliction results from a random and spontaneous mutation of one gene. It also indicates that this mutated gene must be dominant over its normal partner, setting up a cascade of events that accelerates the ageing of the individual.

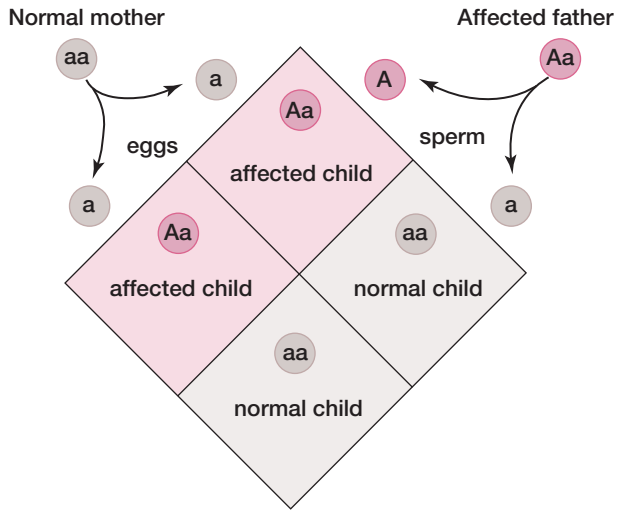


Figure 16.35 One example of autosomal dominant inheritance. Carriers of the dominant allele are affected.

Huntington disease, an autosomal dominant condition, is a lethal disorder in which the brain progressively deteriorates over a period of about 15 years. Its symptoms typically appear after age 35, which is often after the affected individuals have already had children. Early symptoms include irritability and mild memory loss, followed by involuntary arm and leg movements. As the brain deteriorates, these symptoms become more severe, leading to loss of muscular co-ordination, memory, and the ability to speak. Most people die in their forties or fifties without knowing if their children have inherited the mutant allele.

Incomplete Dominance

The disease familial hypercholesterolemia (FH) is caused by incomplete dominance. That is, the heterozygote exhibits a phenotype midway between both dominant and recessive traits. Approximately one in 500 people are heterozygous, inheriting a defective allele for a gene that codes for the production of cell surface proteins called LDL receptors. Circulating LDL (low-density lipoproteins) cholesterols must bind to these receptors in order to be taken up and used by cells. With one defective allele, heterozygotes produce only half the required receptors and exhibit twice the normal blood cholesterol level. Homozygous recessives (about one in 1 000 000 people) do not produce any receptors and can have six times the normal blood cholesterol level. Over time, circulating LDLs build up in artery walls and eventually block them. This causes atherosclerosis, which leads to heart attacks and strokes. While heterozygous individuals may have heart attacks by the age of 35, homozygous recessive individuals can be stricken by a heart attack at the age of two years.

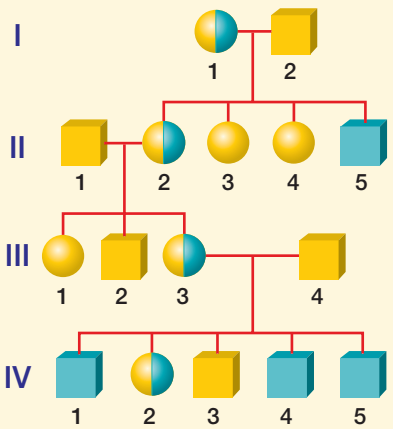
X-Linked Recessive Inheritance

Can you see a number in Figure 16.36? About 8% of men and 0.04% of women cannot identify the number because of colourblindness. Three separate alleles, each coding for a different light-sensitive pigment, are involved in colour vision. The alleles for the pigments that respond to red and green colours are both found on the X chromosome. If one of these two pigments is defective, then the individual cannot distinguish between red and

MINI LAB

Genetic Disorders

This pedigree shows a typical inheritance pattern for Duchenne muscular dystrophy, a disorder that causes the wasting away of muscle tissue. Few individuals with this disorder survive beyond their early 20s. Study the pedigree to determine how the disease is passed from one generation to the next.



Analyze

1. What kind of inheritance does this pedigree indicate?
2. Identify the genotype and phenotype of each individual on the pedigree.
3. How would the pedigree differ if Duchenne muscular dystrophy was an autosomal recessive trait?

green. This condition is known as red-green colourblindness. Why is it so much more common in men than women? The defective allele is a recessive condition. Therefore, for a woman to be colourblind (genotype X^cX^c) her father must be colourblind and her mother must also carry the recessive allele. For a man to be colourblind (genotype X^cY), he only needs to inherit one copy of the recessive allele from his mother. A woman who is heterozygous for the trait (genotype X^CX^c) will not be colourblind, but can pass the gene on to her children.

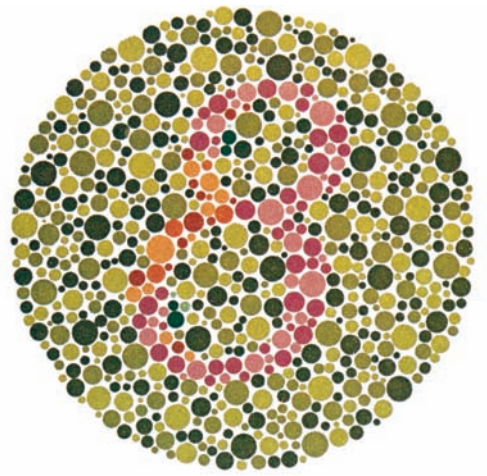


Figure 16.36 People with normal colour vision can determine the number hidden in the dots. Persons with colour blindness cannot distinguish the number.

WEB LINK

www.mcgrawhill.ca/links/atlbiology

The genes responsible for many human genetic disorders have been located on specific chromosomes. The gene associated with PKU, for example, is found on chromosome 12. What other well-known disorders have been mapped to particular chromosomes? What advantages might gene mapping provide to researchers studying inheritance? To find out, go to the web site above, and click on **Web Links**.

Human Genetic Analysis

How do geneticists analyze the pattern of human inheritance? Two techniques that have been used successfully are the examination of karyotypes and the construction of pedigrees.

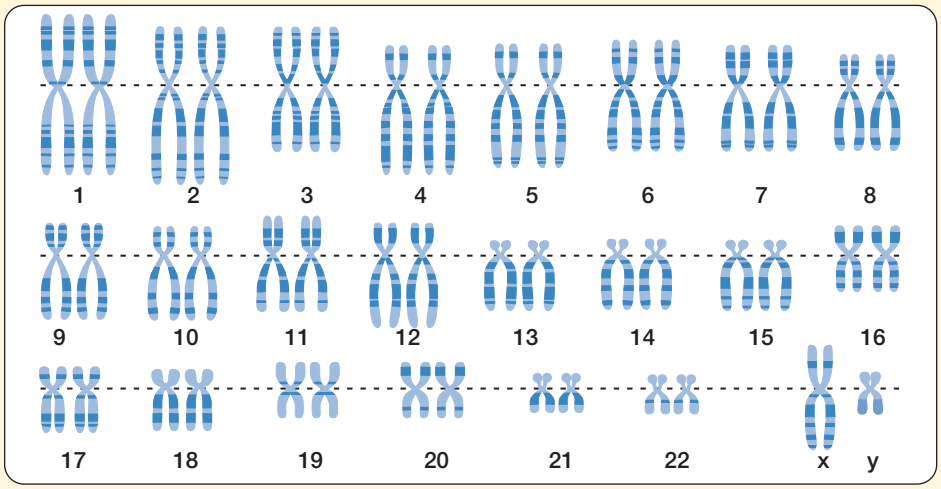
The Human Karyotype

Humans possess 46 chromosomes in every somatic cell; 44 are autosomes and two are sex chromosomes. A **karyotype**, shown in Figure 16.37, is an illustration or photograph of the chromosomes in the nucleus of a somatic cell in an organism. To make a karyotype cells are first grown in a glass container. A special solution is added to stop cell division at metaphase when the chromosomes are clearly visible. Then, the chromosomes are separated from the cells, stained, and photographed.

MINI LAB

What Can a Karyotype Tell Us?

Examine the human karyotype carefully. Count and record the total number of chromosomes in this individual's karyotype, and the number of chromosome pairs. Look at the similarities and differences between the chromosome pairs.



A human karyotype. The chromosomes have been stained to show their bands.

Analyze

1. What characteristics are used to give a chromosome pair a number?
2. Which pair does not have a number? Explain why.
3. What is the sex of the individual? Explain how you know.
4. How would the karyotype differ if this individual had
   (a) Down syndrome?
   (b) Turner syndrome?

Finally, enlarged images of the chromosomes are cut out and arranged in pairs according to their size, shape, and appearance.

Those chromosomes that are difficult to distinguish based on appearance alone are subjected to special staining techniques that reveal the unique banding pattern of each chromosome.

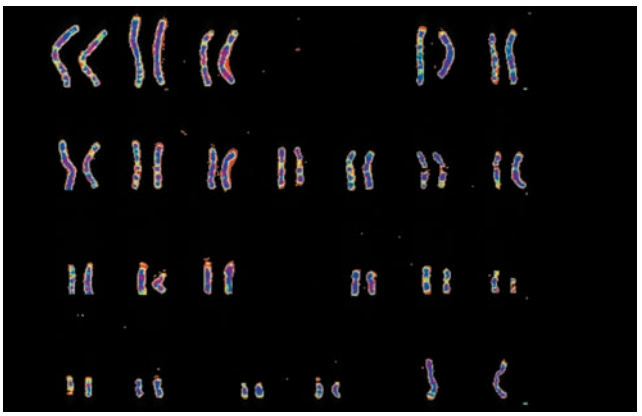


Figure 16.37 This karyotype shows the number of chromosomes found in a normal human body cell. How can karyotypes help to detect genetic disorders?

Constructing Pedigrees

As you saw earlier, a pedigree is a chart that shows the genetic relationships between individuals in a family. After careful data collection involving interviews and direct observations, the pedigree chart is constructed using a standard procedure with specific symbols and definitions. Using a pedigree chart and Mendelian genetics, scientists can determine whether the allele responsible for a given condition is dominant, recessive, autosomal, or sex-linked. Pedigrees can also be used to predict the likelihood of an individual inheriting a genetic disorder, or to deduce the likely genotypes of an affected individual's ancestors and family members.

Hemophilia — which comes from the Latin meaning "loves to bleed" — is a genetic disorder in which the affected person's blood lacks particular clotting factors. As a result, the blood does not clot. This means that even a relatively minor cut or bruise can be life threatening. Hemophilia afflicted some members of Queen Victoria's large family. Examine the pedigree of Queen Victoria and her

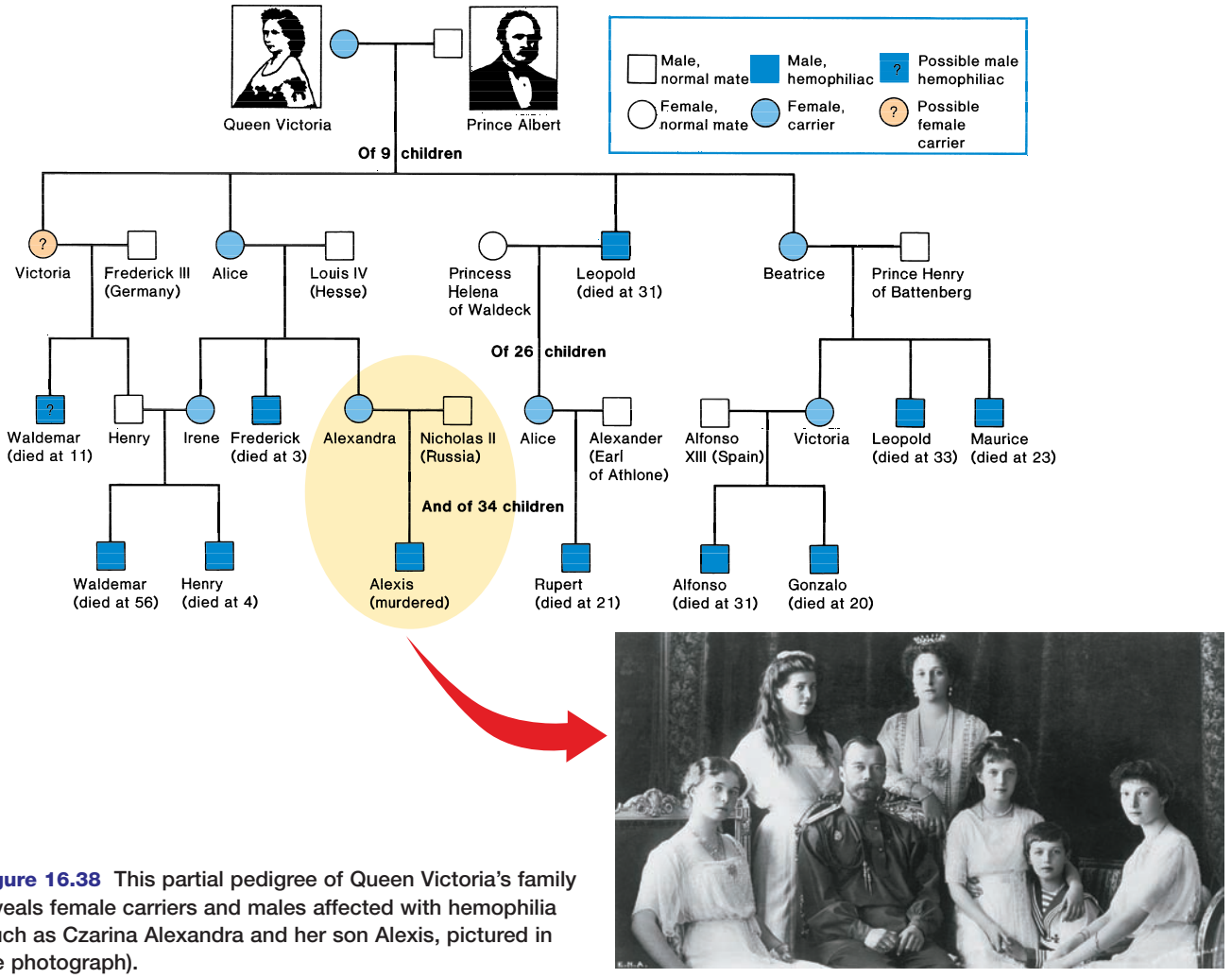


Figure 16.38 This partial pedigree of Queen Victoria's family reveals female carriers and males affected with hemophilia (such as Czarina Alexandra and her son Alexis, pictured in the photograph).

The Case of the Caped Murderer

Background

They found Lord William's body in the library, sprawled on the floor with a dagger in his chest. A copy of his new will, leaving all his money to charity, was found unsigned on his desk. The wealthy Lord William was a bachelor who had no family other than his twelve nephews and nieces. His parents and siblings were all dead.

Inspectors Crick and Watson were the first officers to arrive on the scene. Before becoming a detective, Crick had dabbled in genetics. Glancing at the portrait of Lord William's parents above the fireplace, Inspector Crick explained to those assembled that Lord Edward (William's father) had bright red hair, caused by a recessive pair of alleles, while Lady Iris (his mother) had brunette hair, caused by a dominant allele. Crick further noted that Iris must have been heterozygous, because half of her children had been redheads.

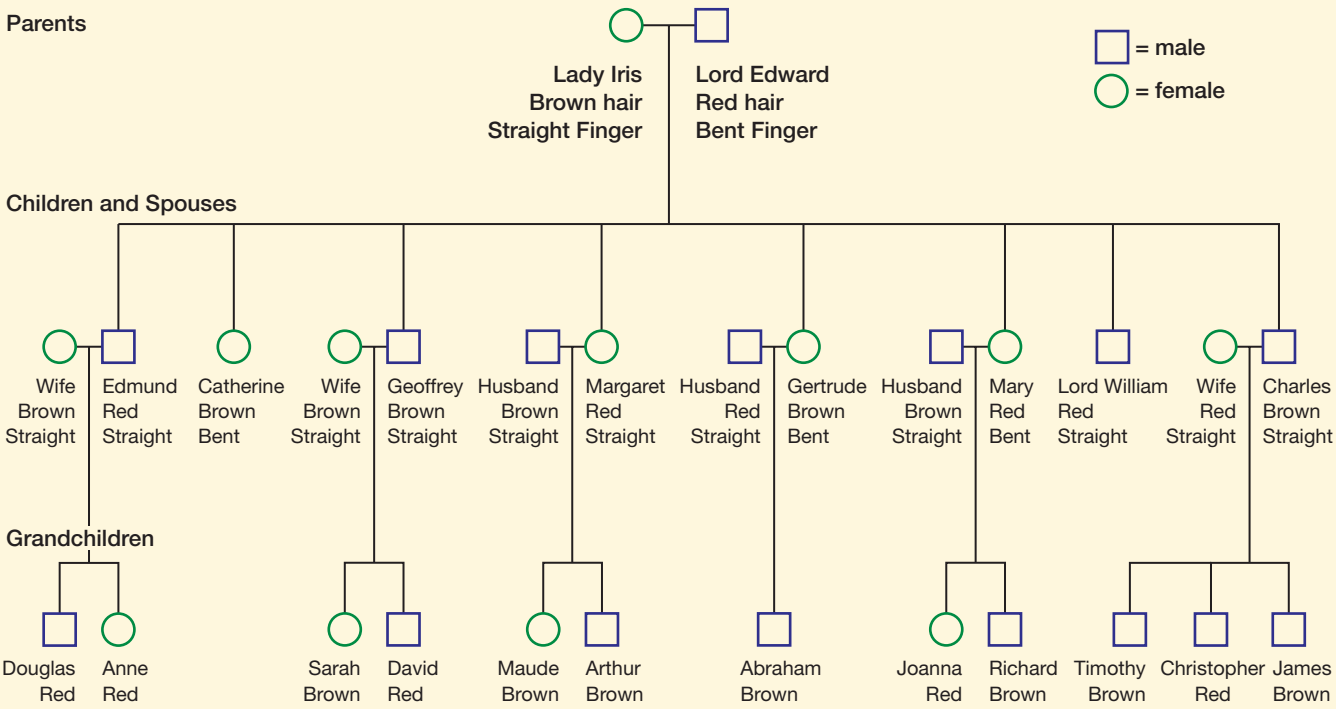
Crick and Watson questioned the family and servants and discovered that there was a witness to the murder. On the fateful evening a maid, upon hearing sounds coming from the library, had spied through the keyhole and seen someone wearing a long, hooded black cape. "I could not tell if it was a man or a woman, sir," she explained, "but I did see red hair sticking out from under the hood. The person was holding a dagger behind his or her back and I noticed that the little finger of that hand was crooked. It was bent inward toward the fourth finger."

"Aha! It's elementary, Inspector Watson!" cried Crick. "A bent little finger is due to a single pair of alleles. If a person carries just one copy of the dominant allele, they will have a bent little finger." Inspector Crick quickly become convinced that the culprit was a niece or nephew who stood to lose an enormous fortune if the new will was signed. To confirm his suspicions, Crick examined old family photographs and portraits, and assembled the following pedigree chart.

You Try It

Study the chart carefully. Determine who has inherited what alleles, and then answer the following questions.

1. Who murdered Lord William?
2. Why didn't Inspector Crick suspect any of Lord William's brothers or sisters?
3. What role did the dominant allele play in unmasking the perpetrator?
4. Why is David not a suspect?



Pedigree of Lord William's family

descendants (see Figure 16.38). How quickly can you detect that males are the ones at risk and that females are the carriers? For female carriers, there is a 50% chance they will pass on the X chromosome that carries the recessive allele to their sons. Since fathers always donate their Y chromosome and not their X chromosome to their sons, a son can only inherit the allele for hemophilia from his mother.

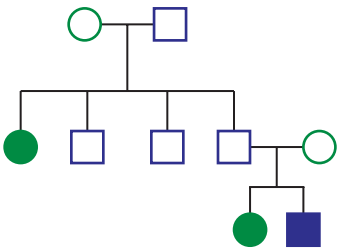
As you saw at the beginning of this chapter, it was not until the late nineteenth century that patterns of inheritance began to be understood. It was even more recently that scientists identified the molecular basis of heredity. In the next chapter, you will learn how molecules in your cells store, transmit, and express genetic information.

BIO FACT

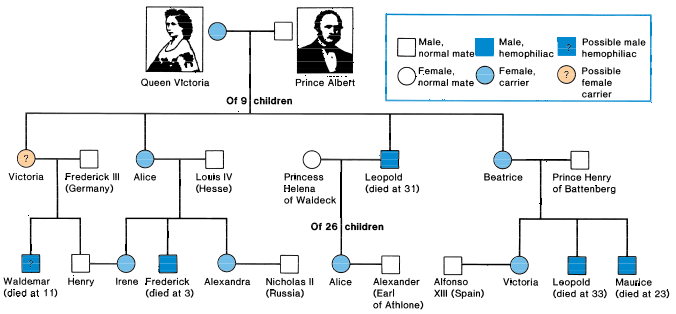
There are no hemophiliacs in the present royal family. Victoria's eldest son, who became King Edward VII, did not inherit the gene and did not marry a carrier. His offspring, therefore, did not inherit the disorder.

SECTION REVIEW

1. Describe how a disorder can be passed along through autosomal recessive inheritance, and explain how this pattern differs from the inheritance of autosomal dominant conditions.
2. What is meant by heterozygote advantage?
3. Can a recessive allele be eliminated from a population? Explain.
4. A colleague tells you, "Men inherit their X chromosome only from their mothers and pass it only to their daughters." Is this statement true? Explain.
5. What steps could you take to start building a list of genetic characteristics of people in your class?
6. Which of the following traits is most likely to be maintained in a population: a fatal autosomal dominant condition that usually develops in individuals before puberty, or the same condition that usually develops in young adults?
7. Which of the following could *not* be determined by observing a human karyotype? Explain.
   (a) the sex of the individual
   (b) the number of homologous pairs of chromosomes
   (c) the presence of Huntington's disease
   (d) the presence of Turner's syndrome
8. What is a karyotype? What type of information can be obtained from it?
9. Indicate how a female could inherit complete red-green colour blindness.
10. Albinism is an autosomal recessive condition. Suppose that George with albinism marries Mary who is not affected. They have a daughter, Frances, who has albinism. Frances marries Jack, whose mother and sister both have albinism. Frances and Jack have a daughter, Anne, who is not affected.
    (a) Construct a pedigree of the family.
    (b) Identify the genotype of each person in the family.
11. The pedigree shown here is of a family carrying the gene for phenylketonuria. Individuals with shaded symbols have the condition.



    (a) How is the disease carried to successive generations?
    (b) In the two generations with PKU, evaluate the parents' roles as carriers. (Are none, one, or both parents carriers?) Explain your answer.
12. This pedigree shows the transmission of hemophilia, an X-linked recessive trait. How would the pedigree differ if
    (a) hemophilia was an autosomal recessive condition?
    (b) hemophilia was a Y-linked trait and it was Prince Albert rather than Queen Victoria who carried the gene?



Chapter Summary

Briefly explain each of the following points.

- A dominant trait is always expressed in an individual. (16.1)
- A homozygous individual is either dominant or recessive for a particular trait. (16.1)
- Punnett squares or the product rule can be used to determine the outcome of crosses. (16.1)
- In pea plants, Mendel found that the inheritance of one trait was not influenced by the inheritance of another trait. (16.2)
- Exceptions to simple Mendelian genetics include incomplete dominance, co-dominance and multiple alleles. (16.2)
- Genes are arranged along chromosomes. (16.3)
- Sex-linked inheritance involves traits carried on genes located on the sex chromosomes (16.3).
- Some traits are determined by more than one gene. (16.3)
- Genetic diseases and disorders can arise from changes in chromosome structure and chromosome number. (16.3)
- The pattern of inheritance of many genetic conditions can be explained by simple Mendelian genetics. (16.4)
- Patterns of human inheritance can be determined through the use of karyotypes and pedigrees. (16.4)

Language of Biology

Write a sentence using each of the following words or terms. Use any six terms in a concept map to show your understanding of how they are related.

- traits
- heredity
- inherited
- genetics
- variations
- purebred
- true breeding
- P generation
- filial generation
- F_1 generation
- hybrid
- monohybrid
- dominant
- recessive
- principle of dominance
- F_2 generation
- Mendelian ratio
- law of segregation
- genes
- alleles
- unit characters
- unit theory
- homozygous
- heterozygous
- probability
- product rule
- Punnett square
- genotype
- phenotype
- complete dominance
- test cross
- dihybrid cross
- law of independent assortment
- incomplete dominance
- co-dominant
- multiple alleles
- pedigree
- carrier
- chromosome theory
- gene-chromosome theory
- sex-linked inheritance
- Barr body
- polygenic inheritance
- continuous variation
- modifier genes
- deletion
- inversion
- duplication
- fragile X syndrome
- translocation
- monosomy
- Down syndrome
- syndrome
- Turner syndrome
- Tay-Sachs disease
- pheylketonuria (PKU)
- albinism
- sickle cell anemia
- heterozygous advantage
- progeria
- Huntington disease
- karyotype

UNDERSTANDING CONCEPTS

1. In your notebook, state whether each of the following statements is true or false. Correct each false statement.

(a) Gregor Mendel is credited with developing the fundamental principles of genetics.

(b) The 1 : 2 : 1 ratio is a ratio of phenotypes in a dihybrid cross.

(c) A hybrid is an organism that is a cross between two different purebred plants.

(d) The second filial generation is the offspring of the parent generation.

(e) The Punnett square is a graphical way of illustrating all the possible combinations of gametes.

(f) The genotype of an organism describes the physical appearance of the particular characteristic under study.

2. Explain the meanings of Mendel's principle of segregation and principle of independent assortment. Under what circumstances does the principle of independent assortment hold true? Give an example in which this law does not apply.

3. What is the difference between a phenotype and a genotype? Does knowledge of an organism's phenotype always lead you to determine the correct genotype? Explain.

4. Explain, using examples, the difference between incomplete dominance and co-dominance.

5. What is the relationship between a chromosome and a gene?

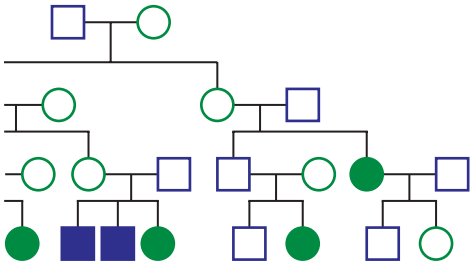
6. One form of baldness is associated with an X-linked recessive trait. Explain why many more men than women are affected by this form of baldness.

7. If Huntington disease is lethal, why does it continue to exist in the population?

INQUIRY

8. In the case of Mendel's pea plants round seeds (R) are dominant over wrinkled seeds (r).
   (a) For which of the plants carrying various combinations of these traits could you tell the genotype by observation alone? Explain.
   (b) Mendel worked out a method for determining the genotype when he could not tell just by looking at the phenotype alone. What was the system he developed? Explain the genotypes he determined using this system.

9. In the pedigree below, white symbols represent individuals who can roll their tongues and the shaded symbols represent individuals who cannot roll their tongues. Determine the genotypes (or possible genotypes) for every individual represented in the pedigree.



10. Your aunt dies and leaves you her mouse Henry. To obtain some money from your inheritance, you decide to breed the mouse and sell the offspring. In looking over Henry's family history, you discover that Henry's grandfather exhibited a rare genetic disorder, which leads to brittle bones. This disorder is hereditary and is the result of being homozygous for the recessive allele (bb). Based on this information, you realize that there is a possibility that Henry could be heterozygous for the allele. If this is the case, the genetic condition may be passed on to his offspring.
    (a) What method could you use to determine if Henry is heterozygous and carries the recessive allele (b)?
    (b) Using the method you described in question (a), determine which phenotypes and genotypes you would expect to see if Henry is heterozygous or if he is homozygous dominant.

11. Show what happens in a cross involving three independent traits at once. Find the ratios of the phenotypes in each case. Black fur (B) is dominant to brown fur (b) in hamsters. Short fur (A) is dominant over long fur (a). Rough coat (R) is dominant over smooth coat (r).
    (a) What happens when a hamster that is homozygous dominant for all three traits is crossed with a hamster that is homozygous recessive for all three traits? Use a Punnett square to determine your answer.
    (b) Determine the genotypes and phenotypes of the offspring of a cross between two organisms from the F_1 generation.

12. Mendel proposed much of his genetic theories based on results he obtained from crossing various pea plants. He crossed a tall pea plant with a short pea plant and observed that all the offspring were tall. From this observation he proposed his law of dominance. How would Mendel's laws have been altered if,
    (a) the results of his cross were all short pea plants?
    (b) the results of his cross were all plants of medium height?
    (c) the results of his cross were plants of many different heights?

13. The F_1 phenytypic ratio for eye colour from a cross between a pair of fruit flies is 1 red eyed female : 1 red eyed male : 1 white eyed male. Determine the genotypes and phenotypes of the parent generation.

14. The F_1 phenotypic ratio for eye colour from a cross between a pair of flies is 2 red eyed females : 2 red eyed males. Determine the genotypes and phenotypes of the parent generation.

COMMUNICATING

15. When we determine genetic ratios that exist among the offspring of different crosses, we are working with probability. Determine the probability of each in the following problems.

(a) A couple are both tested and found to be carriers of the cystic fibrosis gene (cystic fibrosis is a disease caused by the inheritance of two recessive genes). If they have two children, what is the chance that both children will be affected by cystic fibrosis?

(b) What is the chance that both children will be carriers of the trait?

(c) What is the chance that the couple will have two girls who are both affected by cystic fibrosis?

16. The tobacco plant illustrates single gene inheritance. How would you set up an experiment to determine the inheritance of two different characteristics?

17. A wealthy elderly couple dies together in an accident. Soon a man appears to claim their fortune, contending that he is their only child. Other relatives dispute this claim. Hospital records show that the deceased couple had blood types AB and O respectively. The claimant to the fortune is type O. Do you think the claimant is an impostor? Explain your answer.

18. Yellow guinea pigs crossed with white ones always produce cream-coloured offspring. When two cream-coloured guinea pigs are crossed, they produce yellow, cream, and white offspring in the ratio of 1 yellow : 2 cream : 1 white. How are these colours inherited? Explain.

19. Would species that have more traits that are determined through polygenic inheritance than single gene inheritance have a survival advantage over species that have very few traits that are determined through polygenic inheritance? Write a short essay to support and explain your reasoning.

20. Compare and contrast the terms gene and allele.

MAKING CONNECTIONS

21. Mendel chose to conduct his research on the commercial pea plant. What do you think would have happened if he had chosen a different specimen on which to conduct his research?

22. People who are heterozygous for a recessive condition 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? Present your thoughts in a one page essay.

23. Examine the pedigree of the British Royal Family (Figure 16.00 on page 000). How do we determine that it was Queen Victoria and not her husband, Prince Albert, who carried the gene for hemophilia?

24. The IQ (Intelligence Quotient) test was developed in 1903 by Alfred Binet. He believed that IQ could be expressed by taking our mental age, dividing it by our physical age and multiplying by 100. For example, a 10 year old with a mental age of 9 would have an IQ score of 90. Since it was developed, the concept of IQ has been hotly debated. One question is whether there are alleles for IQ. Describe the problems associated with trying to find a scientific explanation for differences in IQ scores.