Patterns of Inheritance

Specific Expectations
In this chapter, you will learn how to . . .
- D1.1 analyze, on the basis of research, some of the social and ethical implications of research in genetics and genomics (5.3)
- D2.3 use the Punnett square method to solve basic genetics problems involving monohybrid crosses, incomplete dominance, codominance, dihybrid crosses, and sex-linked genes (5.2, 5.3)
- D2.4 investigate, through laboratory inquiry or computer simulation, monohybrid and dihybrid crosses, and use the Punnett square method and probability rules to analyze the qualitative and quantitative data and determine the parent genotype (5.2, 5.3)
- D3.2 explain the concepts of DNA, genes, chromosomes, alleles, mitosis, and meiosis, and how they account for the transmission of hereditary characteristics according to Mendelian laws of inheritance (5.1, 5.2, 5.3)
- D3.3 explain the concepts of genotype, phenotype, dominance, incomplete dominance, codominance, recessiveness, and sex linkage according to Mendelian laws of inheritance (5.1, 5.2, 5.3)
- D3.4 describe some genetic disorders caused by chromosomal abnormalities or other genetic mutations in terms of chromosomes affected, physical effects, and treatments (5.3)

Traditionally, controlled breeding of plants that had different desirable traits was used to produce new plants with these combinations. Advances in genetics research now allow more precise and efficient approaches. An example of this transition to more modern practices is the development of Brassica species of plants. Brassica crops are important to Canada’s economy and include canola (B. napus), shown here. In the 1970s, a selective breeding program by Canadian scientists produced a variety of rapeseed (B. campestris) with significantly reduced levels of two toxic chemicals. This new plant was named canola in honour of its Canadian origin (Canadian oil, low acid). Today, one of the largest genetics research programs in Canada involves canola crops.
**Playing the Odds**

Selective breeders rely on patience and keen observation skills to influence the traits of offspring. Modern geneticists rely on the same skills, as well as knowledge from biochemistry, statistical analysis, and other fields of inquiry. Geneticists analyze the data they collect and often use the results to formulate or to test a hypothesis.

How well do you think you can predict results of an experiment based on a hypothesis? How close to the predicted results must the data that you collect be for you to be confident that they support the hypothesis? A coin toss is useful to make and test predictions.

**Materials**
- 2 coins

**Procedure**

1. Working with a partner, determine the probability that any toss of two coins will result in one or both coins being “heads” or neither coins being “heads.” Use a table such as the one below to record your predictions. For 10 pairs of coin tosses, how many “heads” do you expect? How many “no heads” do you expect? Develop a hypothesis that describes these outcomes.

   **Coin Toss Results**

<table>
<thead>
<tr>
<th>Results</th>
<th>“Heads” (Head–Head or Head–Tail)</th>
<th>“No Heads” (Tail–Tail)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Prediction</td>
<td></td>
<td></td>
</tr>
<tr>
<td>(Expected)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Actual</td>
<td></td>
<td></td>
</tr>
<tr>
<td>(Observed)</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

2. You and a partner will each toss a coin. Then you will record the results. Either two heads or a combination of a head and a tail is considered “heads.” Two tails is considered “no heads.”

3. Toss your coins 10 times, and record your results.

**Questions**

1. Calculate your percentage error in the number of “heads” by using the formula below.

   \[
   \text{Percentage error} = \left| \frac{\text{observed} - \text{expected}}{\text{expected}} \right| \times 100\%
   \]

2. Do your data support your hypothesis? Why or why not?

3. Combine the data obtained by the entire class. Calculate the percentage error in the number of “heads.” How does the percentage error for the combined data compare with yours?

4. This activity models how chromosomes are distributed, or segregated, during meiosis. Explain the connection between meiosis and this model.
Numerous plant and animal species that we know today, such as the canola shown in the Chapter Opener and the dog breeds shown in Figure 5.1, are the result of careful and selective breeding. Such breeding was based on the common-sense observation that offspring resemble their parents. People assumed, therefore, that there had to be some way that traits are passed on from parents to the next generation. Certain traits were identified as highly desirable, and breeders used careful decision making and planning so that offspring could have as many of these traits as possible.

Figure 5.1 Beginning with wild canines, such as the Eurasian wolf (A), humans have used selective breeding over centuries to gradually develop numerous breeds of dogs with specific attributes. The Great Pyrenees (B) was bred to protect sheep herds from wolves and bears. The dachshund (C) was bred to hunt badgers in underground dens. The toy poodle (D) was bred to provide a smaller version of the standard poodle, with its intelligence and sensitivity.

Early Ideas About Inheritance

People bred animals and plants for thousands of years without understanding the mechanisms of inheritance. Many people were curious about these mechanisms, however, and tried to explain them.

- The Greek philosopher Aristotle (384–322 B.C.E.) proposed the first widely accepted theory of inheritance, called pangenesís. According to this theory, the egg and sperm consist of particles, called pangenes, from all parts of the body. Upon fertilization of the egg by a sperm, the pangenes develop into the parts of the body from which they were formed. This idea about inheritance was accepted for hundreds of years, even though no experiments were done to test its assumptions or results.

- In 1677, Antony van Leeuwenhoek (1632–1723) discovered living sperm in semen, using a single-lens microscope that he designed. He believed that he saw a complete, miniature person in the head of sperm. Van Leeuwenhoek believed that this miniature person came from the father but developed in the mother.

- During the 1800s, the breeding of ornamental plants became popular. Scientists observed that offspring had characteristics of both parents. The idea of blending became the working theory of inheritance. Scientists thought that characteristics of the parents blended in the offspring. They thought that this blending was irreversible, so that the original characteristics of the parents would not appear in future generations.

These theories were developed to explain different observations that had been made. But none was based on scientific evidence, and they were all eventually disproved over time. It was the scientific evidence of an Austrian monk, Gregor Mendel, that would eventually provide some answers to the question “How are traits inherited?”
Developing a Theory of Inheritance: Gregor Mendel’s Experiments

Great scientific discoveries are sometimes made in the most unexpected places. No one would have expected a great discovery to be made in a monastery garden by an Augustinian monk in Austria, shown in Figure 5.2. The scientists of the time certainly did not expect it and ignored much of this work until other supporting evidence emerged in the 1900s.

Gregor Mendel (1822–1884) studied botany and mathematics at the University of Vienna before entering the monastery. This knowledge proved invaluable for his studies of inheritance. Mendel was successful in sorting out some of the mystery of inheritance in large part because of the plant he chose for his study: the pea plant. Pea plants were available in many varieties and show many traits. In genetics, a trait is a specific characteristic or feature of an organism, such as the flower colour of a plant. The laws that Mendel developed through his work on pea plants formed the foundations of our modern theory of inheritance.

**Mendel’s Pea Plants**

Pea plants reproduce through sexual reproduction, but they usually self-fertilize. This means that the same plant provides both the male and female gametes. The plants that Mendel selected for his study self-fertilized to produce offspring with consistent traits generation after generation. Such plants are called **true breeding**. He obtained his true-breeding plants through selective breeding techniques until he had plants that self-fertilized to produce plants with predictable traits. Mendel also needed to be able to control his experiments. He did this by selectively fertilizing a female gamete with a specific male gamete, in a process called a **cross**. You may also see it referred to as **cross-pollination**.
P generation
breeding, the organisms initially crossed and are typically true breeding
F₁ generation
the offspring of a cross of the P generation
monohybrid cross
a cross of two individuals that differ by one trait

Table 5.1 The Seven Traits of Pea Plants Studied by Mendel

<table>
<thead>
<tr>
<th>Traits</th>
<th>Forms</th>
<th>Traits</th>
<th>Forms</th>
</tr>
</thead>
<tbody>
<tr>
<td>Stem length</td>
<td>tall, short</td>
<td>Flower position</td>
<td>axial, top</td>
</tr>
<tr>
<td>Pod shape</td>
<td>inflated, pinched</td>
<td>Flower colour</td>
<td>purple, white</td>
</tr>
<tr>
<td>Seed colour</td>
<td>yellow, green</td>
<td>Pod colour</td>
<td>green, yellow</td>
</tr>
<tr>
<td>Seed shape</td>
<td>round, wrinkled</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

The Results of Mendel’s True-Breeding Crosses

For his experiments, Mendel chose seven traits that were expressed in two distinguishable forms. These traits and their forms are listed in Table 5.1. Mendel began each experiment with true-breeding plants, which he called the parental or P generation. True-breeding plants with one form of a trait were crossed with true-breeding plants with the other form of the same trait. For example, he crossed true-breeding plants with green-coloured seeds with true-breeding plants with yellow-coloured seeds. Offspring of crosses between plants of the P generation were called the first filial or F₁ generation. This type of cross is called a monohybrid cross because only one trait is monitored in the cross and hybrid plants (those made from parents of differing forms of traits) are produced.

As shown in Figure 5.3, when Mendel grew the seeds produced from a monohybrid cross between yellow-seed plants and green-seed plants, he found that all of the offspring in the F₁ generation had yellow seeds. The green form of seed colour seemed to have disappeared. For all seven of the traits studied, Mendel noticed that when true-breeding organisms with contrasting forms of a trait were crossed, the offspring expressed only one form of that trait.

Figure 5.3 After crossing true-breeding plants, only yellow seeds were produced in the F₁ generation. Note that a cross is represented by the symbol ×.

Identify the form of seed colour that seems to have disappeared. Why could it not really disappear from the pea plant population?
The Results of Mendel’s F₁ Crosses

Mendel also studied pea plants that result from a cross between plants of the F₁ generation. These offspring were called the second filial or F₂ generation. Mendel allowed the plants from the F₁ generation to self-fertilize, and then grew the seeds for the F₂ generation. The results of one of his experiments are shown in Figure 5.4. Based on these results, Mendel realized that the green form of seed colour had not really disappeared in the F₁ generation. Instead, it was unexpressed, since green seed colour could reappear in the F₂ generation. The ratio of plants with yellow seeds to plants with green seeds in the F₂ generation was 6022:2001, or 3.01:1. This is very close to a ratio of 3:1, which is called the Mendelian ratio. For each of the seven traits of pea plants, Mendel discovered that one form of a trait disappeared in the F₁ generation and reappeared in the F₂ generation.

Figure 5.4 For F₂ generation plants, the ratio of plants with yellow seeds to plants with green seeds was approximately 3:1.

The Law of Segregation

Mendel concluded that there must be two hereditary “factors” for each trait he studied. Today, we refer to these factors as alleles. Recall that alleles are different forms of a gene, and that diploid organisms have two alleles for each gene. So, for example, each of Mendel’s pea plants had two alleles for seed colour. For the F₁ generation, although all of the seeds were yellow, they all had a copy of each form of the gene for seed colour. They each had an allele for yellow seeds from one parent, and an allele for green seeds from the other parent. Yellow is the dominant form of seed colour and green is the recessive form of seed colour. Mendel’s work led him to propose the law of segregation. This law states that inherited traits are determined by pairs of “factors,” or two alleles of a gene. These two alleles segregate into each of the gametes of the parents during meiosis, so that each gamete contains one of the alleles. Upon fertilization, each offspring contains one allele from each parent. The form of trait that is expressed in an individual depends on whether they inherit dominant or recessive alleles for the trait. If a dominant allele is present, only the dominant form of the trait will be expressed. Expression of the recessive form requires that an individual has two recessive alleles for the trait.

Learning Check

1. What is a monohybrid cross?
2. In what ratio do traits appear in the F₂ generation when the P generation of plants are true-breeding plants with opposite forms of a trait?
3. How does the F₁ generation from a cross between true-breeding plants with opposite forms of a trait differ from the F₂ generation?
4. Why did Mendel start with true-breeding plants?
5. Round seed shape is dominant to wrinkled seed shape. Draw sketches of monohybrid crosses of P generation and F₁ generation plants that show this.
6. Two brown-eyed parents have two children, one with brown eyes and one with blue eyes. Using the terms dominant and recessive, explain how this can occur.
Genotype and Phenotype

Alleles are often represented using upper-case and lower-case letters. A dominant allele is represented by the upper-case form of the first letter of the allele’s description. The same letter in lower-case is used to represent the recessive allele. The allele for yellow seeds in Mendel’s pea plants is represented by \( Y \), and the allele for green seeds is represented by \( y \). Since every diploid organism has two alleles for each gene, there are three possible allele combinations: two copies of the dominant form, two copies of the recessive form, and one copy of each form. For Mendel’s pea plants, therefore, there are three possible allele combinations for seed colour: \( YY \), \( Yy \), and \( yy \).

As shown in Table 5.2, the combination of alleles an organism has is called the genotype. The expression of a genotype is called the phenotype. A pea plant with a seed colour genotype of \( YY \) would have a phenotype of yellow seeds. Since \( Y \) (yellow) is dominant to \( y \) (green), a pea plant with a genotype of \( Yy \) would also have a phenotype of yellow seeds. Yellow seed colour is referred to as the dominant phenotype. Green seed colour is the recessive phenotype. Only pea plants with a genotype of \( yy \) would have a phenotype of green seeds. An individual with two identical alleles is said to be homozygous for that trait. If the individual has two dominant alleles (\( YY \)), it is homozygous dominant. If the individual has two recessive alleles (\( yy \)), it is homozygous recessive. An individual with two different alleles of a gene (\( Yy \)) is said to be heterozygous.

<table>
<thead>
<tr>
<th>Genotype</th>
<th>Phenotype</th>
</tr>
</thead>
<tbody>
<tr>
<td>YY (homozygous dominant)</td>
<td>Yellow seed</td>
</tr>
<tr>
<td>Yy (heterozygous)</td>
<td>Yellow seed</td>
</tr>
<tr>
<td>yy (homozygous recessive)</td>
<td>Green seed</td>
</tr>
</tbody>
</table>

**Activity 5.1 Tasting Is Genetic**

Specific taste cells in the taste buds on our tongues detect some molecules in the food we eat as bitter. This is because the molecules stimulate specialized proteins on taste cells, called bitter taste receptors. However, some things taste bitter to some people but are tasteless to others. These differences are based on genetics. The genetic link has been known since the 1930s, when it was accidentally discovered that a molecule called phenylthiocarbamide (PTC) tasted very bitter to some people but was tasteless to others. Today, we know that this is due to a gene for a bitter taste receptor and that there is a dominant allele and a recessive allele for this gene.

Before the toxicity of PTC was as well understood as it is today, analyzing for the ability to taste this molecule became one of the first genetic testing methods. This test was even used for paternity testing before DNA testing was available. A representation of data from studies to determine the inheritance of this trait is shown on the right above.

**PTC Testing**

<table>
<thead>
<tr>
<th></th>
<th>Tasters</th>
<th>Non-tasters</th>
</tr>
</thead>
<tbody>
<tr>
<td>Number of individuals</td>
<td>244</td>
<td>81</td>
</tr>
</tbody>
</table>

**Procedure**

Examine the data in the table, and then answer the questions that follow.

**Questions**

1. Based on the data, is the ability to taste PTC a dominant phenotype or a recessive phenotype? Explain.
2. Provide the genotypes for PTC tasters and non-tasters.
3. Many toxins in nature taste bitter. Why are the roughly 30 different genes that humans have for bitter taste receptors an evolutionary advantage?
Section 5.1 REVIEW

Section Summary

- Mendel’s monohybrid cross experiments with pea plants demonstrated the existence of dominant and recessive forms of traits.
- Mendel’s studies led to the law of segregation, which states that alleles of genes separate during meiosis and that each offspring receives one allele from each parent.
- The dominant form of a trait occurs when only one copy of the allele is needed for an individual to express the form. That allele is referred to as the dominant allele. For the recessive form of a trait to be expressed, an individual must have two copies of the recessive allele.
- The combination of alleles in an individual is its genotype. The expression of the genotype is an individual’s phenotype.

Review Questions

1. **K/U** What does it mean to describe an allele as dominant or recessive? Use an example to help you explain your answer.

2. **A** A child has his father’s eyes, his mother’s nose, but his grandmother’s hair colour. The child’s father, the grandmother’s son, does not have the same hair colour as the grandmother. Explain how this combination of traits may be inherited using Mendel’s law of segregation and the principles of dominance and recessiveness.

3. **T/I** The following data were obtained from the initial cross between a true-breeding tall pea plant with a true-breeding short pea plant. Do the data presented in the tables support the Mendelian ratio? Explain your answer, and any differences you observed.

### Results for the F1 Generation

<table>
<thead>
<tr>
<th>Trait</th>
<th>Number of Plants</th>
</tr>
</thead>
<tbody>
<tr>
<td>Tall plants</td>
<td>257</td>
</tr>
<tr>
<td>Short plants</td>
<td>0</td>
</tr>
</tbody>
</table>

### Results for the F2 Generation

<table>
<thead>
<tr>
<th>Trait</th>
<th>Number of Plants</th>
</tr>
</thead>
<tbody>
<tr>
<td>Tall plants</td>
<td>149</td>
</tr>
<tr>
<td>Short plants</td>
<td>53</td>
</tr>
</tbody>
</table>

4. **T/I** A student is attempting to reproduce Mendel’s experiment. For the P generation cross, the student crosses a pea plant that produces yellow peas with a pea plant that produces green peas.

   a. What coloured peas are predicted for the F1 generation plants?
   
   b. The F1 generation resulted in offspring that produced both green and yellow peas. What mistake might the student have made?

5. **T/I** If you did not know that the allele for purple flowers in pea plants was dominant to the allele for white flowers, how could you determine this experimentally?

6. **K/U** Distinguish between the terms phenotype and genotype.

7. **T/I** The term heterozygous can be used on its own, but the term homozygous should be followed by either recessive or dominant. Explain why.

8. **C** Using labelled diagrams, illustrate one of Mendel’s monohybrid cross experiments by following one trait from the P generation to the F2 generation. Include the following labels in your diagrams: genotype, phenotype, heterozygous, homozygous, recessive, and dominant.

9. **K/U** In humans, freckles is dominant to no freckles.

   a. How would you represent the allele for freckles and no freckles?
   
   b. What is the genotype of an individual with no freckles?
   
   c. What is the genotype of an individual with freckles? Can you be sure? Explain.

10. **T/I** Two black guinea pigs produce an offspring with white fur.

    a. What can you infer about coat colour in guinea pigs from this information?
    
    b. What are the possible genotypes of the parents and offspring?

11. **K/U** In fruit flies, the allele for regular wing size is dominant (R) to the allele for vestigial (small, non-functional) wings (r). What is the genotype of each of the following?

    a. a true-breeding fly with regular wing size
    
    b. a fly with vestigial wings
    
    c. a fly that is the offspring of the flies in parts (a) and (b)

12. **K/U** Given that the allele for tall plants (T) is dominant to the allele for short plants (t), describe the phenotypes of the following genotypes.

    a. **TT**  
    b. **Tt**  
    c. **tt**
Since every cell has two alleles for each gene, there are two possible outcomes of meiosis. This is similar to the fact that there are two possible outcomes when you flip a coin. The probability of “heads” when a coin is flipped is $\frac{1}{2}$, just as the probability of a specific allele sorting into a gamete is $\frac{1}{2}$, or 50 percent. Probabilities can be combined. Since the probability in a coin toss of getting “heads” is $\frac{1}{2}$, the probability of the coin landing on heads two times in a row is $\frac{1}{2} \times \frac{1}{2}$, or $\frac{1}{4}$. In other words, there is a 25 percent chance of it occurring.

Analyzing Genetic Crosses: Punnett Squares

A British geneticist named Reginald Punnett (1875–1967) devised a simple, visual technique to help analyze the results of crosses. This technique uses a grid, which is now called the **Punnett square**. A Punnett square for a monohybrid cross is drawn by following the steps outlined in **Figure 5.5**.

Punnett squares use Mendel’s law of segregation to illustrate all of the possible offspring that could be formed from the gametes of the parents. Thus, Punnett squares provide a visual representation of the probability of inheriting a certain genotype and phenotype. There are two possible gametes produced by each parent, each containing one of the alleles. So, the probability of producing either of the gametes is $\frac{1}{2}$. The probability of any particular offspring inheriting a specific combination of the alleles is $\frac{1}{2} \times \frac{1}{2}$, or $\frac{1}{4}$. The four squares in a Punnett square represent the four possible combinations, and each has a $\frac{1}{4}$, or 25 percent, chance of occurring.
A Punnett Square Analysis of Mendel’s Experiments

Figure 5.6 shows a Punnett square that represents a monohybrid cross between individuals in the F₁ generation of Mendel’s pea plant experiment on flower colour. In pea plants, purple flowers are dominant to white flowers. Two heterozygous (Pp) purple-flowered plants in the F₁ generation were created by crossing a true-breeding purple-flowered plant and a true-breeding white-flowered plant. A Punnett square technique is applied to a cross of the F₁ generation, providing a prediction of the phenotypes and genotypes of the F₂ generation.

Figure 5.6 This Punnett square analysis provides the probability of each genotype and phenotype in the F₂ generation. The ratio of phenotypes in the F₂ generation for this example is 3:1. Therefore, it predicts that three purple-flowered plants will form for every one white-flowered plant. This is what Mendel observed.

Test Crosses: Determining the Genotype of a Parent That Has a Dominant Phenotype

When geneticists want to know if an individual is heterozygous or homozygous for a dominant phenotype, they can do a test cross. A test cross is a cross between an individual of unknown genotype for a trait and an individual that is homozygous recessive for that trait. Analyzing the phenotype of the offspring should provide insight into the unknown genotype of the parent. For example, a plant breeder could do a test between a purple-flowered pea plant of unknown genotype (either Pp or PP) and a white-flowered pea plant. By analyzing the phenotype of the offspring, the breeder would gain insight into the genotype of the purple-flowered plant. Figure 5.7 shows the predicted offspring from these two crosses.

Figure 5.7 In a test cross, if any of the offspring show the recessive phenotype, the unknown genotype of the parent must be heterozygous.

**Explain** Even if none of the offspring are white, why can you not be absolutely sure that the genotype of the unknown parent is PP?
Using a Punnett Square to Predict the Genotypes and Phenotypes of Offspring

Problem
In pea plants, the allele for round seeds is dominant to the allele for wrinkled seeds. If a pea plant that is heterozygous for seed shape is crossed with a pea plant that produces wrinkled seeds, determine the following:

a. the genotypes of the P generation plants
b. the genotypes and phenotypes of all of the potential F₁ generation plants
c. the probability of producing offspring with wrinkled seeds in the F₁ generation

What Is Required?
You are asked to find the genotypes of the parents, as well as the predicted genotypes and phenotypes of the offspring. You also need to find the probability of producing offspring with wrinkled seeds.

What Is Given?
One parent is heterozygous, and the other parent produces wrinkled seeds.

Plan Your Strategy
Act on Your Strategy

You are asked to make a cross between two plants. First, determine the genotypes of the two parents.

Since round seeds is dominant to wrinkled seeds, let \( R \) represent the round-seed allele, and \( r \) represent the wrinkled-seed allele. One parent is heterozygous, so that parent's genotype is \( Rr \). The other parent displays the recessive phenotype, so that parent's genotype is \( rr \).

Complete the Punnett square for the cross \( Rr \times rr \).

1. Place the gametes along the top and down one side.

2. Combine the gametes to form the offspring.

Identify the possible genotypes and phenotypes of the offspring.

The predicted offspring produced are:
- 2 \( Rr \) – heterozygous; round phenotype
- 2 \( rr \) – homozygous recessive; wrinkled phenotype

Determine the probability of producing a plant with wrinkled seeds.

The probability of producing a wrinkled-seed plant \( (rr) \) is \( \frac{2}{4} = \frac{1}{2} \) or 50%.

Check Your Solution
Since one parent is heterozygous, \( Rr \), it makes sense that when crossed with a homozygous recessive plant, \( rr \), half of the offspring plants have wrinkled seeds.
Using a Punnett Square to Determine the Unknown Genotype of a Parent

**Problem**
In pigs, the allele for black coat colour is dominant to the allele for white coat colour. Two black pigs mate, producing 5 black piglets and 1 white piglet. Determine the genotypes of the parents. What is the probability of producing a white piglet in the next litter?

**What Is Required?**
You are asked to find the genotypes of the two parents and the probability of producing a white piglet in the next litter.

**What Is Given?**
You know that both parents are black, and that both white and black offspring were produced.

<table>
<thead>
<tr>
<th>Plan Your Strategy</th>
<th>Act on Your Strategy</th>
</tr>
</thead>
<tbody>
<tr>
<td>You are asked to determine the genotypes of the parents. First, determine the genotypes of the offspring.</td>
<td>Since the allele for black coat colour is dominant to the allele for white coat colour, let $B$ represent the black coat colour allele, and let $b$ represent the white coat colour allele. The white piglet must be homozygous recessive, $bb$. The black piglets may be either $BB$ or $Bb$. Since you cannot be sure which genotype is correct, use $B_-$, where the underline character represents a placeholder for either $B$ or $b$.</td>
</tr>
<tr>
<td>Complete what you know in the Punnett square by filling in the genotypes for the known offspring. The common practice is to put the genotype for the recessive phenotype in the bottom right corner. Since you do not know whether the black piglets are $Bb$ or $BB$, put the $B_-$ in the top left corner.</td>
<td>$B_-$</td>
</tr>
<tr>
<td>Since you know that both parents are black, they must be either $Bb$ or $BB$. Since you do not know for sure, put the dominant allele at the left above the squares and at the top left on the side.</td>
<td>$B$ $B_-$</td>
</tr>
<tr>
<td>Now that you have all of the known information in the Punnett square, work backward to complete the Punnett square.</td>
<td>$B$ $BB$ $Bb$</td>
</tr>
<tr>
<td>The genotype of each parent is $Bb$.</td>
<td>$b$ $Bb$ $bb$</td>
</tr>
<tr>
<td>Determine the probability of a white piglet from the Punnett square.</td>
<td>The probability of a white piglet is $\frac{1}{4}$, or 25%.</td>
</tr>
</tbody>
</table>

**Check Your Solution**
Since two black-coated parents produce an offspring with a recessive phenotype (white coat colour), each must have a recessive allele. The probability of a white piglet in the next litter is based on how many times $bb$ appears in the Punnett square, which is 1 out of the 4, or 25%.
Practice Problems

1. Determine the gametes produced by the following parents, given that axial flowers are dominant to terminal flowers in pea plants.
   a. AA    c. homozygous recessive
   b. Aa    d. heterozygous

2. The allele for purple flowers (P) is dominant to the allele for white flowers (p). Predict the genotypes and phenotypes of the offspring produced from a cross between a plant with the genotype Pp and a plant with the genotype pp.

3. Green pod colour (G) is dominant to yellow pod colour (g) in pea plants. Predict the phenotypes and genotypes of the offspring produced when a plant homozygous for green pods is crossed with a plant homozygous for yellow pods.

4. The allele for long wings is dominant to the allele for curly wings in fruit flies. Predict the genotypes and phenotypes of the offspring resulting from a cross between two fruit flies heterozygous for wing shape.

5. The allele for tall pea plants is dominant to the allele for short pea plants. What is the probability that two plants, heterozygous for height, will produce a plant heterozygous for height?

6. The allele for short beaks is dominant to the allele for long beaks in a particular species of bird. Show all the different crosses that result in 50 percent of the offspring having long beaks.

7. In Alaskan malamute dogs, the allele for dwarfism is recessive (n) to the allele for normal-sized dogs (N). Two normal-sized dogs have three puppies—two are normal sized and one is dwarf sized.
   a. What is the genotype of each parent?
   b. What are the probabilities of the genotypes and phenotypes for puppies in the next litter from the same parents?

8. In mice, a condition called waltzer is recessive. A waltzer mouse (nn) has a defect of the inner ear that interferes with balance and causes the mouse to run in circles. A mouse that runs normally is crossed with a waltzer mouse. Half the offspring are waltzer mice. What are the genotypes of the parents?

9. Two pigs of unknown genotype produce a litter of piglets, six black and one white. If the allele for white coat colour is recessive to the allele for black coat colour, determine the genotypes of the parents, if possible. If it is not possible, what other information do you need?

10. Two plants were crossed, producing all blue flowers in the F1 generation. When one of the plants was allowed to self-fertilize, the ratio of flowers produced by the plants was 3:1 blue to yellow. Since the allele for blue flowers is dominant to the allele for yellow flowers, determine the likely genotype of the P generation.

Learning Check

7. List all the possible different gametes that could be produced from the following parents:
   a. Tt
   b. GG
   c. ff

8. What is the predicted phenotypic ratio of offspring from a cross between two individuals who are heterozygous for a trait?

9. Look at the data in Figure 5.5. What is the predicted phenotypic ratio of offspring from this cross, if R represents round seeds and r represents wrinkled seeds?

10. Draw an example of a Punnett square that shows a monohybrid cross between an individual who is heterozygous for a trait and another individual who is homozygous recessive for the same trait.

11. The allele for round seeds is dominant to the allele for wrinkled seeds in pea plants. Two pea plants, heterozygous for seed shape, are crossed to produce 280 offspring. What is the expected number of round-seeded plants and wrinkle-seeded plants?

12. In humans, dimpled cheeks is dominant to non-dimpled cheeks. Is it possible that two dimpled adults have a non-dimpled child? Explain.
**The Inheritance of Two Traits: Dihybrid Crosses**

Having determined the inheritance pattern of one trait in pea plants, Mendel designed a second set of experiments that involved following two traits. In these experiments, Mendel set out to determine whether the pattern of inheritance for one trait, such as plant height, had any impact on the pattern of inheritance for another trait, such as flower colour. This time, Mendel started with plants that were true bred for two traits, and each parent differed in both of the traits. Since two traits are involved, this type of cross is called a **dihybrid cross**.

In one of his experiments, Mendel crossed true-breeding plants that produced yellow, round seeds (**YYRR**) with true-breeding plants that produced green, wrinkled seeds (**yyrr**). Mendel discovered that all the **F**\(_1\) generation plants displayed the dominant forms of each trait—they all produced yellow, round seeds. This is the same result he obtained when following only one trait in monohybrid crosses. When Mendel then let the plants self-fertilize, as he had in his monohybrid crosses, the recessive forms of each trait reappeared in the **F**\(_2\) generation. **Figure 5.8** summarizes what Mendel observed.

![Diagram of a dihybrid cross](image)

**Figure 5.8** One of Mendel’s dihybrid crosses involved true-breeding pea plants with yellow, round seeds and true-breeding pea plants with green, wrinkled seeds. In the **F**\(_1\) generation, the ratio of plants with yellow, round seeds to plants with yellow, wrinkled seeds, to plants with green, round seeds to plants with green, wrinkled seeds was 315:101:108:32, which is close to 9:3:3:1. **Identify the genotypic ratio for each of the traits independently.**

**Developing the Law of Independent Assortment**

Mendel performed and repeated numerous dihybrid crosses. He discovered that no matter which two traits he followed, he always obtained the same ratio of 9:3:3:1. That is, for every one offspring displaying recessive forms of both traits (**yyrr**), there would be three offspring displaying one recessive form of a trait and a dominant form of the second trait (**Y_rr**), three offspring displaying the opposite combination of recessive and dominant forms of the two traits, (**yyR_**), and nine displaying the dominant forms of both traits (**Y_R_**).
A Punnett Square Can Model Mendel’s Results

Figure 5.9 shows how a Punnett square can be used to model the segregation of the alleles for one of Mendel’s dihybrid cross experiments. In the F\textsubscript{2} generation, each of the parents could produce four possible gametes (YR, Yr, yR, and yr), since each of the alleles for colour could combine with each of the alleles for shape. When the four different gametes from one parent combine with the four different gametes from the other parent, 16 possible genotypes are produced. These are the typical 16 squares in the Punnett square for a dihybrid cross. The resulting possible phenotypes include:

- 9 displaying the dominant forms of both traits
- 3 displaying one dominant and one recessive form of each trait
- 3 displaying the other dominant/recessive pair of traits
- 1 displaying both recessive forms of each trait

These are exactly the same ratios that Mendel obtained experimentally. These are expected only if the inheritance of one trait has no influence on the inheritance of another trait. Mendel described these events in the **law of independent assortment**. Using current terminology, this law states that the alleles for one gene segregate or assort independently of the alleles for other genes during gamete formation.

**Figure 5.9** In the F\textsubscript{2} generation, individuals in the largest group (9) have at least one dominant allele for each gene (Y\textsubscript{r}R\textsubscript{r}). In the intermediate groups (3), the individuals have at least one dominant allele for one gene, but two recessive alleles for the other gene (Y\textsubscript{r}rr or yyR\textsubscript{r}). The smallest group is homozygous recessive for both genes (yyrr).

<table>
<thead>
<tr>
<th>Genotype</th>
<th>Phenotype</th>
<th>Number</th>
<th>Phenotypic Ratio</th>
</tr>
</thead>
<tbody>
<tr>
<td>Y\textsubscript{<em>}R\textsubscript{</em>}</td>
<td>yellow round</td>
<td>315</td>
<td>9:16</td>
</tr>
<tr>
<td>yyR\textsubscript{_}</td>
<td>green round</td>
<td>108</td>
<td>3:16</td>
</tr>
<tr>
<td>Y\textsubscript{r}rr</td>
<td>yellow wrinkled</td>
<td>101</td>
<td>3:16</td>
</tr>
<tr>
<td>yyrr</td>
<td>green wrinkled</td>
<td>32</td>
<td>1:16</td>
</tr>
</tbody>
</table>
Learning Check

13. What is the difference between a dihybrid cross and a monohybrid cross?

14. List all the possible different gametes that could be produced from parents with the following genotypes.
   a. TtGg  
   b. AABbcc

15. Why might it be important to understand the inheritance pattern of more than one gene at a time?

16. A tall true-breeding plant with purple flowers is crossed with a short true-breeding plant with white flowers. Assuming tall and purple are dominant phenotypes, what is the predicted phenotypic ratio of the offspring?

Sample Problem

Using a Punnett Square to Analyze a Dihybrid Cross

Problem
In wolves, the allele for brown eyes is dominant to the allele for blue eyes. Also, the allele for grey coat is dominant to the allele for black coat. Predict the genotypes and phenotypes of the potential offspring of a blue-eyed grey wolf that had a black-coated mother and a wolf that was heterozygous for both traits.

What Is Required?
You are asked to predict the genotypes and phenotypes of the offspring of the two wolves.

What Is Given?
You know that the allele for a grey coat is dominant to the allele for a black coat, and that the allele for brown eyes is dominant to the allele for blue eyes. You also know that one wolf is heterozygous for both genes, and that the other wolf has a grey coat and blue eyes, and had a black-coated mother.

<table>
<thead>
<tr>
<th>Plan Your Strategy</th>
<th>Act on Your Strategy</th>
</tr>
</thead>
</table>
| You are asked to predict the genotypes and phenotypes of the offspring. First determine the genotypes of the two parents from the given information. | Since grey coat is dominant to black coat, let \( G \) = grey and \( g \) = black. Since brown eyes is dominant to blue eyes, let \( B \) = brown and \( b \) = blue. Since the grey wolf (\( G_\)_) had a black-coated mother, it must be heterozygous for coat colour, having received a \( g \) allele from the mother (\( gg \)) and a \( G \) allele from the father. Therefore,  
   • blue-eyed grey wolf = \( bbGg \)  
   • heterozygous wolf = \( BbGg \)  
| Determine the gametes that each of the parents can produce. | • blue-eyed grey wolf = \( bG, bg \)  
   • heterozygous wolf = \( BG, Bg, bG, bg \) |
| Place the gametes of each parent along the top and side of a Punnett square and complete the square to determine the genotypes and phenotypes of the offspring. In this case, the Punnett square is only eight squares because there are only two possible types of gametes for the blue-eyed grey wolf. | There are six different genotypes: \( BbGG, BbGg, Bbgg, bbGG, bbGg, \) and \( bbgg \). The wolves may be grey with brown eyes (\( BbGG, BbGg \)), black with brown eyes (\( Bbgg \)), grey with blue eyes (\( bbGG, bbGg \)), or black with blue eyes (\( bbgg \)). |
| Examine the results from the Punnett square. |  |

Check Your Solution
Since the genotypes of the parents are \( bbGg \) and \( BbGg \), it makes sense that six different genotypes will be observed.
11. A short pea plant that is heterozygous for flower colour is crossed with a homozygous tall pea plant that is also heterozygous for flower colour.
   a. Determine the genotypes of the plants.
   b. What gametes can the plants produce?

12. There are two genes involved in the length and colour of the coat in guinea pigs:
   • One gene controls the coat colour, which can be either black or brown.
   • The other gene controls coat hair length, which can be either long or short.
When a black long-haired guinea pig was mated with a brown short-haired guinea pig, only black short-haired guinea pigs were produced. Which alleles are dominant and which alleles are recessive? Use genotypes to explain how the parents produced only black short-haired guinea pigs.

13. Summer squash (Cucurbita pepo) come in different varieties. They may be either white (dominant) or yellow (recessive), and may be disk-shaped (dominant) or sphere-shaped (recessive). Determine the probability of producing a yellow disk-shaped squash from a cross between two squash that are heterozygous for both traits.

14. The long hair of Persian cats is recessive to the short hair of Siamese cats, but the black coat colour of Persians is dominant to the black-and-tan coat of the Siamese.
   a. If a true-breeding black long-haired Persian is mated with a true-breeding black-and-tan short-haired Siamese, what are the predicted phenotypes of the cats in the F₁ generation?
   b. If two of the F₁ generation cats are then mated, what is the chance of obtaining a long-haired black-and-tan cat in the F₂ generation?

15. In humans, dark hair is dominant to blond hair, and curly hair is dominant to straight hair.
   a. What are the predicted phenotypes of the children of a man with straight hair homozygous for dark hair and a woman with blond hair who is heterozygous for curly hair?
   b. How would we know that the parents were heterozygous for these traits?

16. A test cross was performed on a pea plant to determine its genotype. The offspring from the test cross were 50 percent green, round seeds and 50 percent green, wrinkled seeds. What is the genotype of the pea plant in question?

17. The allele for dry earwax (w) is recessive to the allele for wet earwax (W), and the allele for attached earlobes (u) is recessive to the allele for unattached earlobes (U). Determine the probability of producing an offspring with the genotype WwUu from two individuals heterozygous for both traits.

18. Two pea plants produce the following offspring:
   • 225 tall with purple flowers
   • 250 short with white flowers
   • 237 tall with white flowers
   • 241 short with purple flowers
Which of the following parental pairs is more likely—two plants heterozygous for both genes, or one heterozygous for both genes and one homozygous recessive for both genes?

19. Two curly haired parents with cleft chins have three children. One child has curly hair and a smooth chin, while the other two children have straight hair and cleft chins.
   a. Is straight hair or curly hair the dominant phenotype? Explain.
   b. Is a cleft chin or a smooth chin the dominant phenotype?
   c. Determine the genotypes of the parents.
   d. If these parents have another child, what is the probability that the child will have straight hair and no cleft chin?

20. Some alleles are lethal when two copies are present. One such allele is the creeper allele, C.
   • Normal chickens are homozygous recessive for the creeper allele.
   • Creeper chickens are heterozygous, Cc, and are born with stunted legs.
   • Chickens homozygous for the creeper allele die.
   • In poultry, feathered legs are dominant to clean legs.
What is the probability of a normal chicken with feathered legs being born from a cross between two creeper chickens that are heterozygous for feathered legs? Hint: Only live births count in the probability ratio.
The Chromosome Theory of Inheritance

When Mendel performed his pea plant experiments in the mid-1800s and formulated his laws of inheritance, he had no idea how the traits were passed from generation to generation. The process of meiosis and the existence of chromosomes had not been discovered. Beginning in the early 1900s, scientists began to take note of similarities between how Mendel's traits segregated and assorted and how chromosomes behaved during meiosis.

Sutton Links Mendel's Work to Chromosome Segregation

In 1902, Walter Sutton (1877–1916) was working as a graduate student at Columbia University in New York. As part of his work, he studied the process of synapsis (segregation of homologous chromosomes) and migration of sister chromatids during meiosis I and meiosis II. Sutton realized that the behaviour of chromosomes during meiosis was related to the behaviour of the factors in Mendel's experiments with pea plants. Expressed from today's perspective and using current terminology, Sutton realized that the distribution of chromosomes into developing gametes follows the pattern for two alleles of a gene, according to Mendel's law of segregation, shown in Figure 5.10. During gamete formation, alleles segregate just as homologous chromosomes do. Sutton proposed that genes are carried on chromosomes, and his work formed the basis for the chromosome theory of inheritance. This theory states that genes are located on chromosomes, and chromosomes provide the basis for the segregation and independent assortment of alleles.

![Figure 5.10](image.png)

Figure 5.10  Alleles (or Mendel’s factors) and chromosomes both segregate during meiosis. During anaphase I, the homologous chromosomes segregate and move to opposite ends of the cell. After telophase I, the homologous chromosomes are in separate cells. The resulting gametes are equally likely to contain each possible combination of alleles.

Infer what would happen to two alleles for different genes that are carried on the same chromosome. Would they most likely be inherited together or assort independently?
Section Summary

- Punnett squares can be used to analyze the results of a genetic cross to predict the probabilities of offspring genotypes and phenotypes. They can also be used to determine an unknown genotype of a parent, typically performed as a test cross.
- The law of independent assortment states that for the inheritance of two traits the alleles for the two associated genes assort independently. The inheritance of one trait does not affect the inheritance of another trait.
- A dihybrid cross of individuals heterozygous for both traits yields offspring with a predicted phenotypic ratio of 9:3:3:1.
- The chromosome theory of inheritance states that genes are located on chromosomes, and that chromosomes provide the basis for the segregation and independent assortment of alleles.

Review Questions

1. Using an illustration, explain how a Punnett square can be used to model a monohybrid cross.

2. The allele for black hair \((B)\) is dominant to the allele for white hair \((b)\) in guinea pigs. Assume that two heterozygous guinea pigs are crossed.
   a. Write the genotypes of the two parents.
   b. Use a Punnett square to predict the genotypes of the offspring.
   c. What is the probability of producing an offspring with white hair?

3. Curly hair is dominant to straight hair in humans. Is it possible for a curly haired man to father curly haired children if his wife has straight hair? Explain using a Punnett square.

4. What is the purpose of a test cross?

5. For prize-winning animals that are used for stud (that is, they are bred to produce offspring that could also win prizes), the breeder must be certain of the animal's genotype. In pigs, the allele for black coat colour is dominant to the allele for white coat colour. How can a breeder be certain of the genotype of a black pig that is being considered for stud?

6. Why does a Punnett square that is used for tracking a dihybrid cross between two heterozygotes have 16 squares?

7. What is the law of independent assortment? Illustrate your answer with an example.

8. Tall stems and purple flower colour are dominant in pea plants. A pea plant is heterozygous for height and flower colour.
   a. Describe the phenotype of the pea plant, given the information in Table 5.1.
   b. Provide the genotype of the pea plant.
   c. List the genotypes of the gametes it produces.

9. In tomato plants, red fruit \((R)\) is dominant to yellow fruit \((r)\), and tall \((T)\) is dominant to short \((t)\). True-breeding tall plants that produce red fruit were crossed with true-breeding short plants that produce yellow fruit.
   a. Predict the genotypes and phenotypes of the \(F_1\) generation plants.
   b. List the genotypes of the gametes produced by the \(F_1\) generation plants.
   c. Predict the genotypes and phenotypes of the \(F_2\) generation plants. Include the genotypic and phenotypic ratios of the \(F_2\) generation plants.

10. In corn, the allele for purple kernels is dominant to the allele for yellow kernels, and smooth texture is dominant to wrinkled. Determine the likely genotypes of the parents if offspring consisted of 46 wrinkled purple kernels, 54 smooth yellow kernels, 153 smooth purple kernels, and 15 wrinkled yellow kernels.

11. In fruit flies, long wings are dominant to short wings. Also, grey body colour is dominant to black body colour. Flies that are heterozygous for both traits are crossed, and 256 offspring are produced. Predict how many of the offspring would be long-winged and grey, long-winged and black, short-winged and grey, and short-winged and black.

12. Explain how the chromosome theory of inheritance supports Mendel's laws of segregation and independent assortment.
Controlled breeding of plants and animals has been the basis for many developments and breakthroughs in genetics research. However, breeding experiments are not ethical, or even possible, for human genetics. For example, researchers cannot perform test crosses between selected men and women. As well, researchers cannot accumulate large numbers of offspring from the same parents to improve the statistical reliability of their data. Instead, human geneticists collect as much information as they can and use it to create a diagram called a pedigree. A **pedigree** is a type of flowchart that uses symbols to show the inheritance patterns of traits in a family over many generations. Figure 5.11 shows an example of a pedigree, as well as the meanings of the symbols used. If you have ever seen a family tree, you may notice some similarities. Family trees and pedigrees use many of the same symbols.

In common terms, a pedigree is a way to analyze how a trait “runs in the family.” Geneticists can use a pedigree to determine the pattern of inheritance of a particular trait that is controlled by a single gene or more than one gene. Pedigrees help uncover the genotype of a particular member of a family and—with a Punnett square analysis—the probability of a particular genotype or phenotype in an offspring. Many pedigree analyses are used to determine the presence or absence of an allele that is responsible for a disease in an individual.

**Key Terms**
- **pedigree**
- autosomal inheritance
- autosomal dominant
- autosomal recessive
- genetic counsellor
- gene therapy

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**Figure 5.11** A pedigree is used to illustrate the inheritance pattern of a particular trait in a family. Standard symbols are used to indicate what is known about the trait being studied. The term **affected** means an individual who has or shows a particular trait. These individuals are indicated in a pedigree by darkened or coloured symbols.
Autosomal Inheritance

As you learned in Chapter 4, autosomes are any chromosomes other than sex chromosomes. **Autosomal inheritance** refers to the inheritance of traits whose genes are found on the autosomes—that is, chromosomes 1 to 22 in humans.

All of the human traits you have learned about so far, such as curly or straight hair and freckled or non-freckled skin, are due to autosomal genes. For each of these genes, one allele is dominant to the other. Autosomal genes are also responsible for many inherited genetic disorders. These disorders may be classified as either autosomal dominant or autosomal recessive. An **autosomal dominant** disorder occurs when the disease-causing allele is dominant and an individual has one or both copies of the allele. An **autosomal recessive** disorder occurs when the disease-causing allele is recessive and an individual has both copies of the allele. Table 5.3 describes some examples of human autosomal genetic conditions. Notice how these diseases can show inheritance patterns that are either autosomal dominant or autosomal recessive.

**Table 5.3** Examples of Human Diseases with Autosomal Inheritance Patterns

<table>
<thead>
<tr>
<th>Chromosome Number</th>
<th>Condition</th>
<th>Inheritance Pattern</th>
<th>Description</th>
</tr>
</thead>
</table>
| 4                 | Huntington disease                | autosomal dominant  | • neurological disease that results in a loss of muscle control and decline in mental ability  
• decreased life expectancy                                                                 |
| 7                 | cystic fibrosis                   | autosomal recessive | • causes thick mucus to build up in the lungs, making breathing difficult and leading to infection  
• blocks the pancreas, stopping digestive enzymes from reaching the intestines  
• decreased life expectancy                                                                                                                                 |
| 11                | sickle cell anemia *See page 244* | autosomal recessive | • red blood cells are irregularly shaped  
• caused by an abnormal hemoglobin protein  
• decreased life expectancy                                                                                                                                 |
| 12                | phenylketonuria (PKU)             | autosomal recessive | • prevents the breakdown of phenylalanine, leading to developmental delays in cognitive function  
• must be treated immediately to avoid symptom development                                                                                              |
| 13                | retinoblastoma                    | autosomal dominant  | • tumours develop in the retina of young children  
• fatal if not treated                                                                                                                                 |
| 15                | Marfan syndrome                  | autosomal dominant  | • affects connective tissue, leading to weakness in the heart, blood vessels, and skeleton  
• causes very long limbs  
• increased susceptibility to heart and blood vessel conditions                                                                                         |
| 15                | Tay-Sachs disease                 | autosomal recessive | • progressive destruction of the nervous system caused by a lack of the enzyme hexosaminidase A, leading to the accumulation of lipids in the cells  
• fatal in early childhood                                                                                                                                 |
| 18                | Niemann-Pick disease              | autosomal recessive | • brain and nervous system impairment due to the accumulation of lipids in cells  
• decreased life span  
• Type A fatal in early childhood                                                                                                                                 |
| 19                | maple syrup urine disease         | autosomal recessive | • inability to break down three amino acids, leading to nerve degeneration  
• fatal if not treated                                                                                                                                 |
| 20                | adenosine deaminase deficiency    | autosomal recessive | • deficiency in the enzyme adenosine deaminase resulting in minimal immune response and susceptibility to all diseases  
• fatal if not treated with bone marrow transplant                                                                                                       |
**Autosomal Dominant Inheritance**
By studying a pedigree, you can determine whether the inheritance pattern of a disorder is autosomal dominant or autosomal recessive. For example, examine the pattern of inheritance in Figure 5.12. In this pattern of inheritance, the child is not affected, and both parents are. This occurs for autosomal dominant disorders and if both parents are heterozygous.

![Figure 5.12](image)

**Figure 5.12** An unaffected child born of two affected parents indicates autosomal dominant inheritance.

**Huntington Disease, an Autosomal Dominant Disorder**
The pedigree shown in Figure 5.13 traces Huntington disease in a family. Huntington disease is a genetic disorder that has an autosomal dominant inheritance pattern. It is a lethal disorder in which the brain deteriorates over a period of about 15 years. In most affected people, symptoms include decreased muscle coordination, erratic body movements, personality changes, and a decline in mental abilities. There is no cure for Huntington disease, but medications can lessen some of the symptoms.

Because Huntington disease is autosomal dominant, all affected individuals must have at least one copy of the dominant allele, \( A \). Therefore, all of the individuals not affected by the disease have the genotype \( aa \). For a family such as the one outlined in Figure 5.13, children of individual II-5 and II-6 have the disorder in their family. However, they have no risk of developing the disease themselves, because both of their parents are homozygous recessive. As a result, all of their children will be disease-free as well. For children of II-1 (\( aa \)) and II-2 (\( Aa \)), there is a 50 percent chance that they will develop Huntington disease.

![Figure 5.13](image)

**Figure 5.13** This pedigree shows the inheritance pattern for an autosomal dominant disorder. Notice that an affected child must have at least one affected parent to be affected, and that once a trait is no longer in the family, it will not reappear.

*Explain why individual II-2 must be heterozygous.*
**Autosomal Recessive Inheritance**

As shown in Figure 5.14, the pattern of inheritance for autosomal recessive disorders differs from the pattern for autosomal dominant disorders. In this pattern of inheritance, two unaffected parents can have an affected child. This can happen only if both parents are *carriers*. Individuals are carriers when they are heterozygous. They *carry* the recessive allele for the trait, but do not express it. Therefore, the individuals are considered to not be affected by the genetic disorder.

![Figure 5.14](image)

**Figure 5.14** In autosomal recessive inheritance, if both parents are heterozygous for the disorder, they will have an affected child.

**Cystic Fibrosis, an Autosomal Recessive Disorder**

The pedigree shown in Figure 5.15 traces cystic fibrosis in a family. Cystic fibrosis is the most common fatal genetic disorder affecting young Canadians. The symptoms of cystic fibrosis are caused by a defective protein that disrupts the movement of chloride ions across cell membranes. The flow of chloride ions controls the movement of water into and out of cells. The water is needed to produce thin mucus that flows freely and easily. Mucus acts as a protective barrier against infection and helps keep membranes moist. However, the thickened mucus that occurs with cystic fibrosis builds up in the organs of the body, causing breathing and digestive problems, and proneness to infections. There is no cure for cystic fibrosis. However, improvements in treating the associated symptoms have increased the life expectancy of people with the disease. In the 1960s people with cystic fibrosis rarely lived beyond 5 years of age. Currently, the average life expectancy is in the late 40s.

Since cystic fibrosis is an autosomal recessive disorder, only homozygous recessive individuals are affected. Notice that parents who do not show symptoms of cystic fibrosis could have an affected child if both parents are heterozygous. Such parents are carriers of the cystic fibrosis gene.

![Figure 5.15](image)

**Figure 5.15** This pedigree shows the inheritance pattern for an autosomal recessive disorder. Notice that the appearance of the recessive phenotype can skip generations, and that two unaffected parents can have an affected child.
Although most traits in humans are influenced by more than one gene, some are controlled by single genes. Examples of single-gene traits and their different forms are shown below.

**Procedure**
1. Work with a partner. Choose a single-gene human trait from the examples provided. Then, choose a family to collect data from. This can be your family or your partner’s family. Alternatively, your teacher can provide you with data.
2. Interview family members, including grandparents, aunts, uncles, and cousins. Find out which phenotype each person exhibits for your chosen trait.
3. Using the symbols in Figure 5.11, create a pedigree for the trait.

**Questions**
1. From the pedigree, determine whether the phenotype is due to a dominant or recessive allele.
2. Fill in as much of each genotype as you can with the data you have. To complete this, fill in genotypes based on the phenotypes. Depending on the data you have, use Punnett squares to work backward to solve for any genotypes of parents and offspring.
3. Do all the data fit the pattern you might expect? If not, what factors could explain the difference?

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**Learning Check**

17. Describe and draw the following pedigree symbols.
   a. male
   b. affected female
   c. mating
18. What does the term *autosomal inheritance* mean?
19. Draw a pedigree showing the mating between an affected man and a woman to produce three children, two unaffected boys and one affected girl. Label the generations and the individuals.
20. What is the key indicator of autosomal recessive inheritance in a pedigree like the one shown in Figure 5.15?
21. A certain autosomal recessive genetic disorder is lethal in children. Explain how this allele can continue to appear in the population even though children with the disease do not survive long enough to have offspring of their own.
22. In certain families in Norway, woolly hair (hair that looks like sheep’s wool) is inherited. In order for children to have this phenotype, at least one of their parents must have woolly hair. Is woolly hair an autosomal dominant or autosomal recessive inherited phenotype? Draw a pedigree for a family in which one of three children and both parents have woolly hair. Identify the genotypes for each individual. Whose genotype can you not be certain of?
Genetic Tests

There are many methods for testing individuals who are at risk of developing genetic disorders, or of passing these disorders on to their offspring. Karyotyping is one method, which you learned about in Chapter 4. It can be used to identify some chromosomal abnormalities, such as extra or missing chromosomes, and abnormal banding patterns. Once geneticists are able to identify the chromosome and eventually the gene responsible for a disorder, more detailed analyses are possible. Some of the types of genetic tests that are available are summarized in Table 5.4. Note that genetic tests are not just for inherited genetic disorders. They are also used to detect chromosomal disorders, such as those that were discussed in Chapter 4.

Table 5.4 Types of Genetic Tests

<table>
<thead>
<tr>
<th>Method</th>
<th>What Is Analyzed</th>
<th>Example</th>
</tr>
</thead>
<tbody>
<tr>
<td>Karyotype</td>
<td>• chromosome structure and number</td>
<td>• Down syndrome: extra copy of chromosome 21</td>
</tr>
<tr>
<td>FISH (fluorescence in situ hybridization)</td>
<td>• details of chromosomal abnormalities; is based on visualizing, through fluorescence, a targeted region on a chromosome</td>
<td>• chronic myelogenous leukemia (CML): part of chromosome 9 has broken off and attached to chromosome 22</td>
</tr>
<tr>
<td>Gene testing</td>
<td>• mutation(s) in the DNA sequence of a gene</td>
<td>• breast cancer susceptibility gene T and 2 (BRCA1 and BRCA2): a woman’s chance of developing breast and/or ovarian cancer is greatly increased if she inherits one or more mutated forms of the gene(s)</td>
</tr>
<tr>
<td>Biochemical testing</td>
<td>• abnormal enzymes and other proteins (often due to mutation of a gene that codes for the protein)</td>
<td>• Tay-Sachs disease: majority of cases are due to lack of activity of the hexosaminidase A protein</td>
</tr>
</tbody>
</table>

Genetic Testing for Cystic Fibrosis and Huntington Disease

In 1989, the gene for cystic fibrosis was identified in the laboratory of Lap-Chee Tsui, shown in Figure 5.16, at the Hospital for Sick Children in Toronto. The gene was determined to be on chromosome 7 and was named CFTR (cystic fibrosis transmembrane conductance regulator). Once the gene was identified, scientists began working to develop a genetic test. Although all forms of cystic fibrosis are caused by a mutation in the CFTR gene, there are over 1600 different mutations. As a result, available tests are able to successfully identify the presence of a mutated CFTR gene about 85 to 90 percent of the time. Approximately one in every 3600 children born in Canada has cystic fibrosis, and one in every 25 Canadians is a carrier for the defective allele. Cystic fibrosis is much more common in Caucasians than in other ethnic groups.

In 1983, the gene for Huntington disease, called huntingtin, was the first human disease-associated gene to be mapped to a chromosome. Ten years after being mapped to chromosome 4, the DNA sequence of the gene was determined. The huntingtin gene contains a region where a CAG nucleotide sequence is repeated several times. The number of times that this sequence is repeated indicates whether an individual will have Huntington disease. The current genetic test for Huntington disease is designed to determine the number of these CAG repeats in an individual’s huntingtin gene. Individuals who have fewer than 35 repeats will not be affected. Individuals with 36 to 39 repeats are at risk of developing the disease, and those with more than this number of repeats will definitely develop the disease. The number of repeats may increase or decrease between generations, and a greater number of repeats is associated with increased severity or earlier onset of the disease.

Figure 5.16 The cystic fibrosis gene was identified in Lap-Chee Tsui’s laboratory at the Hospital for Sick Children.
**Genetic Counselling**

When there is a history of a genetic disorder in a family, the family is often referred to a genetic counsellor. Genetic counsellors have specialized training in human genetics and in counselling individuals and families who may have or be at risk for a genetic disorder. When working with a family, as shown in Figure 5.17, a genetic counsellor can use a pedigree to determine the genotypes of the family members in order to counsel them on options for genetic testing and to explain the probability of passing on a disease-causing allele to their children. Genetic counsellors also assist individuals with genetic disorders that are not inherited.

Genetic counsellors can explain the symptoms of genetic disorders and the available treatments, provide other information, and, equally importantly, provide emotional support. For example, individuals who have tested positive for mutations of the *BRCA1* or *BRCA2* genes that cause an increased risk of breast and ovarian cancers can receive counselling for deciding what to do next. Genetic counselling is also available for families expecting a new baby who have decided on or are considering prenatal genetic testing.

**Decisions About Genetic Testing**

There are many issues related to genetic testing and what to do with the results once they are known. Some issues have family-related implications. Decisions may need to be made about who will provide long-term care to someone who has a disorder. Also, there may be some feelings of guilt felt by members of the family who test negative for a genetic disorder. Although understanding the inheritance of a disorder cannot help individuals who are already affected, it can help families make decisions about having additional children. The significant difference between Huntington disease and other genetic disorders is the time of onset. Because those who have Huntington disease do not usually display any symptoms until after age 35, they may have already had children when they are first diagnosed. There are also work-related issues. Individuals worry that being diagnosed with a terminal illness may affect their job opportunities, their ability to get medical insurance, or the way they are treated at work.

If there is no cure for the disorder, some people may prefer not to know that they may be affected, while others may want to plan for the future. In some cases, such as positive tests for *BRCA1* and *BRCA2*, identification of the mutated gene does not guarantee that the individual will experience the disease. In addition, early detection of some disorders can open up additional treatment options that would not be available once symptoms appear.
An incorrect DNA sequence of a gene can cause a genetic disorder in humans. The cell starts producing the protein that is coded by this gene, thereby correcting the genetic disorder.

1. An incorrect DNA sequence of a gene can cause a genetic disorder in humans.
2. A copy of the correct DNA sequence for a gene is placed inside a piece of viral DNA that acts as a carrier.
3. The virus with the correct gene is taken up by the human cells. A copy of the correct gene is now present in the human cells.
4. The cell starts producing the protein that is coded by this gene, thereby correcting the genetic disorder.

Gene Therapy: A Cure for Genetic Disorders?

Although genetic testing provides options to those carrying an allele that is associated with a genetic disorder, the greatest hope for those living with genetic disorders may lie with gene therapy. Gene therapy is a technique aimed at correcting the effects of a mutated gene that is associated with a genetic disorder by inserting the correct form of the gene into the genome of the patient. Thus, the goal is for gene therapy to be able to cure all genetic disorders.

In gene therapy, a copy of the normal gene is inserted into a vector, which acts as a carrier of the gene. The vector is usually the DNA of a virus, because viruses can easily infect human cells. Once the virus delivers the normal gene to the human cell, the correct protein that is made from the gene is produced. The basic principles of gene therapy are shown in Figure 5.18.

Figure 5.18 In gene therapy, the aim is to insert a copy of a gene into the genome of a patient. The new gene produces the correct protein and corrects the genetic disorder.

The Future of Gene Therapy

Researchers have been working on gene therapy techniques since the 1990s. However, gene therapy is still in the experimental stage due to two significant obstacles: the type of vector that is used and poor integration of the gene into chromosomes. Although viruses are effective vectors, they present the potential of an immune response. Also, benefits of gene therapy continue to be short-lived. Although the new genes get transferred to human cells, they are not successfully integrated into the cells’ chromosomes. Therefore, the normal gene does not get reproduced along with the rest of the cell’s genes during cell division.

Scientists continue to work on safer and more effective mechanisms of gene transfer. Research has demonstrated that a number of genetic disorders, including cystic fibrosis and Huntington disease, can be successfully treated with gene therapy once these barriers have been overcome. In addition, researchers are also investigating the use of gene therapy to treat other types of illnesses, such as heart disease. In 2000, researchers at St. Michael’s Hospital in Toronto performed Canada’s first human gene therapy treatment for heart disease. They introduced a gene into a patient’s heart tissue that produces a protein that stimulates growth of new blood vessels and increased blood flow in the heart.
Section Summary

- A pedigree is a key tool for geneticists who study the inheritance of human traits.
- A pedigree can be used to determine the inheritance pattern of a trait (autosomal dominant or autosomal recessive) and provide information about the genotypes and phenotypes of previous and future generations.
- Genetic testing is used to detect changes to chromosome structure or number, as well as disease-causing genes. Karyotyping, fluorescent in situ hybridization (FISH), and gene testing are three methods that are used.
- Gene therapy, although still in the experimental stage, holds great promise for curing genetic disorders by modifying the genetic information in cells.

Review Questions

1. **K/U** List three uses for a pedigree.
2. **T/I** Which type of inheritance pattern, autosomal dominant or autosomal recessive, is likely to remain hidden in a family, only to appear in a future generation? Explain your answer.
3. **K/U** Determine the pattern of autosomal inheritance if two affected parents have an unaffected child.
4. **K/U** What significant contribution to cystic fibrosis research was made in Ontario?
5. **A** A woman had cystic fibrosis in her family and did not want to have a child who suffered from the disease. She and her spouse went for genetic testing and counselling. She tested positive as a carrier and her spouse tested negative, and their first child was born with cystic fibrosis. Explain how this could happen.
6. **C** Using a graphic organizer, summarize ethical and social concerns associated with people being tested for a genetic disorder. Include the pros and the cons for testing.
7. **T/I** Examine the pedigree below.
   - a. Determine whether the inherited trait displayed is autosomal dominant or autosomal recessive.
   - b. Provide the genotypes of all of the individuals in the pedigree. If there are any genotypes that you cannot be certain of, explain why.

8. **T/I** Students were given the pedigree below and asked to identify the inheritance pattern. One student thought it represented autosomal recessiveness and another student thought it represented autosomal dominance. A third student argued that it could represent either pattern of inheritance. Who is correct? Explain why.

9. **C** Construct a pedigree using the following information. Albinism, a lack of skin pigmentation, is a recessive phenotype. An individual with this disorder is referred to as albino. Two parents with normal pigmentation have four children, three girls and a boy. The eldest girl and the boy are both albino. Determine the genotypes of all of the members of the family. For those whose genotype cannot be determined, what is the probability that they are heterozygous? (Hint: Since you know they are not albino, do not count that possibility in your calculation.)

10. **T/I** There are many forms of testing that can be used to identify the presence of a genetic condition. Differentiate between the types of testing according to how they can be used.

11. **K/U** Describe three roles a genetic counsellor may play in helping a family cope with the possibility of a genetic disease.

12. **A** How does gene therapy compare with other cloning techniques discussed in this unit?

13. **T/I** “Gene therapy will soon be able to provide a cure for all genetic disorders.” Do you agree or disagree with this statement? Provide a reason for your response.
Monohybrid Crosses in *Brassica rapa*

In this investigation, you will work with plants called Wisconsin Fast Plants™ (*Brassica rapa*). They have been designed to germinate and mature quickly. Your goal is to collect evidence to explain how stem colour in *Brassica rapa* is inherited. The trait you are focussing on, stem colour, is controlled by a gene that regulates whether anthocyanin (a purple pigment) is expressed.

The seeds used to begin the investigation are produced from the parental cross between a true-breeding purple-stemmed plant and a true-breeding green-stemmed plant. For the investigation, the seeds from the F<sub>1</sub> generation are grown and the stem colours of the plants recorded. Then the plants from the F<sub>1</sub> generation are pollinated and the seeds harvested and used to grow the F<sub>2</sub> generation plants. Stem colours for that generation of plants are also recorded. To save time, your teacher may perform some of these steps for you.

**Pre-Lab Questions**

1. How does this investigation differ from Mendel's first experiments?
2. When crossing two true-breeding plants, what phenotype do you expect to be displayed in the F<sub>1</sub> generation?
3. What ratio of phenotypes do you expect in the F<sub>2</sub> generation?

**Materials**

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

**Safety Precautions**

- Wash your hands after handling any of the materials in this investigation.

Go to Organizing Data in a Table in Appendix A for help with designing tables for data.

These *Brassica rapa* plants are shown at (A) 48 hours after seed planting, (B) 3 days growth, and (C) full growth at around 14 days.
Question
How is stem colour inherited in *Brassica rapa* plants?

Procedure

Part I: The *F*₁ Generation
1. Use the instructions provided by your teacher to germinate the seeds and tend to the developing plants.
2. After about four days, observe the stem colour and all other phenotypes of the young *F*₁ plants. Record your observations in a suitable table.
3. Tend to the *F*₁ plants by thinning them to two per pot. Refer to the instructions provided by your teacher.
4. Before beginning Part II, complete the Analyze and Interpret section of the investigation for Part I.

Part II: The *F*₂ Generation
5. After about a week and a half, pollinate the *F*₁ plants over three days. Be sure that all the flowers receive pollen from several different plants. (Refer to the instructions provided by your teacher.)
6. Over the next several days, cut off any new flower buds that developed after you pollinated the plants. Let them dry for a full week.
7. After about two weeks, stop watering the plants. Let them dry for a full week.
8. After about one week, harvest the seeds from the pods of the *F*₁ plants. (Refer to the instructions.) These are your seeds for the *F*₂ generation. Plant the seeds.
9. After about four days, observe the stem colour and other phenotypes of the young *F*₂ plants. Design a table to record your observations.
10. Return all the plants and other materials to your teacher.

Analyze and Interpret

Part I
1. From the data you recorded on the appearance of the stems in the *F*₁ generation, which stem colour is dominant—purple or green? Explain your answer.
2. Given your response to question 1, form a hypothesis about the phenotypic ratio that you will observe in the *F*₂ generation.

Part II
3. Why are the new flower buds cut off the plant in Procedure step 6?

Conclude and Communicate

4. Examine your hypothesis and your results. How well did your results confirm your hypothesis? Identify any sources of error.
5. Draw two Punnett squares illustrating the *P*, *F*₁, and *F*₂ generations to summarize the results of your investigation.

Extend Further

6. **INQUIRY** What other plant traits can be traced using Mendel’s experimental process? Choose one of these traits and design a procedure to investigate its expression in *Brassica rapa*.

7. **RESEARCH** Dr. Paul Williams, a professor in Plant Pathology at the University of Wisconsin and a graduate of the University of British Columbia, developed the *Brassica rapa* strain that you used in this investigation. Along with being rapid-growing, what other traits have been selectively bred into *Brassica rapa* to make it an ideal plant for genetic study?
Corn Genetics

Did you know that when you look at an ear of corn, each one of those kernels is actually an offspring with its own unique genetics? In this investigation, you will examine the pattern of inheritance of one trait in corn—kernel colour. The allele for purple kernels is dominant to the allele for yellow kernels. By counting the number of purple and yellow kernels produced in the F\textsuperscript{2} generation, you can verify the pattern of inheritance for kernel colour.

Pre-Lab Questions

1. Describe the genotypes of the P generation plants that would be used to study the pattern of inheritance for kernel colour.
2. What kernel colour is displayed in the F\textsuperscript{1} generation?
3. What phenotypic ratio is expected in the F\textsuperscript{2} generation?

Question

What is the predicted phenotypic ratio for kernel colour in the F\textsuperscript{2} generation?

Organize the Data

1. Design a table to record the number of purple and yellow kernels in the ear of corn.
2. Count the number of purple and yellow kernels, and record the results in the table.

Analyze and Interpret

1. Determine the ratio of purple to yellow kernels in the ear of corn.

Conclude and Communicate

2. How does the actual ratio of purple to yellow kernels compare to the theoretical ratio?
3. Is the actual ratio close enough to the theoretical ratio to confirm that the allele for purple kernel colour is dominant to the allele for yellow kernel colour? Explain any differences.

Extend Further

4. INQUIRY What improvement could be made to the procedure to better replicate the Mendelian ratio that is expected?
5. RESEARCH Corn, also known as maize, has been used as a model to study genetic processes for over 75 years. The work of Dr. Barbara McClintock is one important example of such a use. Research this scientist and the highly significant scientific contributions that she made to the field of genetics.
Dihybrid Crosses

In this investigation, you will model Mendel’s experiment of dihybrid crosses, which eventually led to the development of the law of independent assortment. You will do this by following two single-gene traits in *Drosophila melanogaster*, the common fruit fly.

**Pre-Lab Questions**
1. Describe the genotypes and phenotypes of the P generation that Mendel used for his dihybrid cross experiment with pea plant seeds.
2. What traits were observed in the F₁ generation of Mendel's experiment?
3. What phenotypic ratio was observed in the F₂ generation of Mendel’s experiment?

**Organize the Data**
1. Choose two traits from the table on the left to investigate.
2. Determine the genotypes of the flies for the P generation.
3. Use a computer simulation or obtain results for the F₁ generation from your teacher. Record the results in a table.
4. Determine the genotypes of the flies for the F₁ cross.
5. Use a computer simulation or obtain results for the F₂ generation from your teacher. Record the results in a table.

**Analyze and Interpret**
1. From the data you recorded for the F₁ generation, which phenotypes are dominant? Explain your answer.
2. What is the expected phenotypic ratio for the F₂ generation?
3. Calculate an actual phenotypic ratio for your results of the F₂ generation.

**Conclude and Communicate**
4. Describe the inheritance pattern for the two traits you investigated.
5. How does the actual phenotypic ratio compare to the theoretical phenotypic ratio? Account for any differences.

**Extend Further**

6. **INQUIRY** In this investigation, you studied the inheritance pattern of two traits. Design an experiment to study the inheritance of all four traits. What results do you expect?
7. **RESEARCH** *Drosophila melanogaster* is one of the most studied animal models in genetics. What makes them useful for such studies? What other organisms are extensively used for genetics research?
To meet the 40-hour community involvement requirement for your graduation diploma, you are volunteering at a local hospital. One of your first assignments is in a clinic for people who have genetic disorders. The patients and their families come to the clinic to meet with their doctors and evaluate their treatment. A key role of this clinic is to facilitate meetings between patients and physicians who are investigating alternative treatments for genetic disorders. One treatment that physicians are researching is gene therapy.

Many patients are nervous when they come to the clinic. They have questions and concerns, especially about new experimental techniques. The hospital has developed a website (below) that describes the science of gene therapy. However, some people are concerned about the clinic’s research into gene therapy and have developed a handout to publicize their concerns. (See page 233.)

**Gene Therapy**

**Weighing the Risks and Benefits**

**Scenario**

To meet the 40-hour community involvement requirement for your graduation diploma, you are volunteering at a local hospital. One of your first assignments is in a clinic for people who have genetic disorders. The patients and their families come to the clinic to meet with their doctors and evaluate their treatment. A key role of this clinic is to facilitate meetings between patients and physicians who are investigating alternative treatments for genetic disorders. One treatment that physicians are researching is gene therapy.

Many patients are nervous when they come to the clinic. They have questions and concerns, especially about new experimental techniques. The hospital has developed a website (below) that describes the science of gene therapy. However, some people are concerned about the clinic’s research into gene therapy and have developed a handout to publicize their concerns. (See page 233.)

**The Science of Gene Therapy**

- Each of our cells contains genetic material, in the form of DNA. DNA carries the information needed for us to stay alive, and it is "passed on" or inherited by our children. The functional units of DNA are called genes. Genes carry the information needed to produce molecules called proteins. All of the processes in the body depend on proteins.
- If a gene becomes damaged or does not work properly, the protein that is made from it also becomes damaged, or is not produced at all. This can cause health problems in the form of a genetic disorder.
- Gene therapy involves inserting a corrected form of a damaged gene into an individual’s DNA. A vector is used as a carrier of the gene, which is needed to insert new DNA into the cell’s DNA. The new gene will then begin making the correct protein, thus curing the genetic disorder.
- Gene therapy can be administered in two ways. The new gene may be inserted into a person’s sperm or egg. The genetic material of that person’s children and all subsequent generations will also have that new gene. Alternatively, the new gene may be inserted into any other cell of the body. In that case, only the person undergoing treatment is affected.
- Gene therapy offers great potential for people with debilitating or fatal conditions such as Huntington disease, Parkinson’s disease, cystic fibrosis, and Duchenne muscular dystrophy.
Gene Therapy: Genetic Engineering of People!

• Genetic engineering involves changing an organism’s genetic material. Gene therapy is a form of genetic engineering that is applied to humans.

• A commonly used vector is a virus. There is no guarantee that the virus will find the correct point in the person’s DNA. If the gene is inserted in the wrong place, it will cause further errors in the genetic material. This could result in even worse consequences and the individual becoming even more ill.

• The immune system of the person undergoing treatment may attack and destroy the virus. In some cases, patients may need to go through many gene therapy sessions. This may cause the immune system to develop more and stronger responses to the virus.

• Gene therapy opens the door for many disturbing applications of genetics. The potential to use gene therapy so that all subsequent generations are affected is a way of making “designer babies.” The gap between wanting a healthy child, free of a genetic disorder, and wanting to design a child to be a musical or mathematical genius is not very wide.

• Some people feel that meddling with the fundamental structure that gives each person his or her uniqueness is equivalent to “playing God.”

Dr. A Delgado
for the concerned members of our community

Research and Analyze

1. In several clinical studies, gene therapy has been linked to cancer and other fatal conditions. Research the side effects of gene therapy, and design a table that summarizes the therapeutic risks and benefits. Use this to perform a risk–benefit analysis of gene therapy. Refer to Analyzing STSE Issues in Appendix A for help with performing this analysis.

2. Scientists at the McEwen Centre for Regenerative Medicine in Toronto have developed a gene therapy technique to repair donor lungs before they are transplanted. The technique dramatically improves the success of transplants. As a result, more donor lungs are available to people who are waiting on transplant lists. Research this technique. Decide, from an ethics standpoint, whether this type of gene therapy is in the same category as treating the actual genetic material of an individual. Write a paragraph that summarizes your opinion, including supporting details.

3. Compare gene therapy with using gene cloning to produce transgenic organisms. Summarize the similarities and differences between the two technologies. As part of your analysis, assess the relative success of the applications and whether one poses more ethical dilemmas than the other.

Take Action

1. Plan In a group, discuss the ethics of gene therapy. What are the differing points of view about using gene therapy? What key issues must be considered when a person is deciding whether to undergo gene therapy? Share the results of the research and analysis you conducted in questions 1 to 3 above.

2. Act Prepare a draft of an information pamphlet that could be checked carefully by medical experts and then handed out to the patients and families visiting the gene therapy clinic. Make sure that your pamphlet helps people understand possible consequences of the decisions they will be making. Support the information you include in your pamphlet with footnotes and a bibliography to show your sources. Ensure that your sources are credible.
### Section 5.1 Understanding Inheritance

Inherited traits can have dominant and recessive forms. The form of a trait that is expressed depends on the two associated alleles that are inherited by an individual.

**KEY TERMS**
- cross
- dominant
- F<sub>1</sub> generation
- F<sub>2</sub> generation
- genotype
- heterozygous
- homozygous
- law of segregation
- monohybrid cross
- P generation
- phenotype
- recessive
- trait
- true breeding

**KEY CONCEPTS**
- Mendel's monohybrid cross experiments with pea plants demonstrated the existence of dominant and recessive forms of traits.
- Mendel's studies led to the law of segregation, which states that alleles of genes separate during meiosis and that each offspring receives one allele from each parent.
- The dominant form of a trait occurs when only one copy of the allele is needed for an individual to express the form. That allele is referred to as the dominant allele. For the recessive form of a trait to be expressed, an individual must have two copies of the recessive allele.
- The combination of alleles in an individual is its genotype. The expression of a genotype is an individual's phenotype.

### Section 5.2 Studying Genetic Crosses

Punnett squares can be used to study the probability of genotypes and phenotypes in monohybrid and dihybrid crosses.

**KEY TERMS**
- chromosome theory of inheritance
- dihybrid cross
- law of independent assortment
- Punnett square
- test cross

**KEY CONCEPTS**
- Punnett squares can be used to analyze the results of a genetic cross to predict the probabilities of offspring genotypes and phenotypes. They can also be used to determine an unknown genotype of a parent, typically performed as a test cross.
- The law of independent assortment states that for the inheritance of two traits the alleles for the two associated genes assort independently. The inheritance of one trait does not affect the inheritance of another trait.
- A dihybrid cross of individuals heterozygous for both traits yields offspring with a predicted phenotypic ratio of 9:3:3:1.
- The chromosome theory of inheritance states that genes are located on chromosomes, and that chromosomes provide the basis for the segregation and independent assortment of alleles.

### Section 5.3 Following Patterns of Inheritance in Humans

Pedigrees can be used to study inheritance patterns in humans. It is hoped that gene therapy will one day be used to cure genetic disorders.

**KEY TERMS**
- autosomal dominant
- autosomal inheritance
- autosomal recessive
- gene therapy
- genetic counsellor
- pedigree

**KEY CONCEPTS**
- A pedigree is a key tool for geneticists who study the inheritance of human traits.
- A pedigree can be used to determine the inheritance pattern of a trait (autosomal dominant or autosomal recessive) and provide information about the genotypes and phenotypes of previous and future generations.
- Genetic testing is used to detect changes to chromosome structure or number, as well as disease-causing genes. Karyotyping, fluorescent in situ hybridization (FISH), and gene testing are three methods that are used.
- Gene therapy, although still in the experimental stage, holds great promise of curing genetic disorders by modifying the genetic information in cells.
Knowledge and Understanding

Select the letter of the best answer below.

1. Purple flowers are dominant to white flowers in pea plants. Which of the following statements about a cross between a true-breeding purple-flowered plant and a true-breeding white-flowered plant is true?
   a. The F1 offspring will be light purple.
   b. The recessive phenotype is expressed in the F1 generation.
   c. The ratio of purple to white flowers in the F1 generation is 3:1.
   d. The offspring in the F1 generation are heterozygous.
   e. The dominant phenotype will not appear in the F2 generation.

2. Brown hair is dominant to blond hair in humans. What is the probability of a heterozygous brown-haired man and a blond-haired woman producing a blond-haired child?
   a. 100%
   b. 75%
   c. 50%
   d. 25%
   e. 0%

3. The allele for freckles is dominant to the allele for no freckles. A freckled man and an unfreckled woman have two children, a boy with freckles and a girl without freckles. What are the genotypes of all four?
   a. Mom: ff, Dad: FF, boy: Ff, girl: ff
   b. Mom: ff, Dad: Ff, boy: Ff, girl: ff
   c. Mom: FF, Dad: ff, boy: Ff, girl: FF
   d. Mom: ff, Dad: FF or Ff, boy: Ff, girl: Ff
   e. Mom: FF or Ff, Dad: ff, boy: Ff, girl: Ff

4. In pea plants, the allele for tall plants is dominant to the allele for short plants, and the allele for yellow pea colour is dominant to the allele for green pea colour. What is the genotype of a short pea plant produced from a cross between a true-breeding yellow pea plant and a true-breeding green pea plant?
   a. TTYY
   b. TYYy
   c. ttyy
   d. ttYY
   e. TtYy

5. Which of the following connects the principles of independent assortment of alleles and the segregation of chromosomes during meiosis?
   a. the chromosome theory of inheritance
   b. the principle of dominance
   c. the law of segregation
   d. the law of independent assortment
   e. the dihybrid cross

6. Which of the following terms can be used to describe a “carrier”?
   a. homozygous dominant
   b. homozygous recessive
   c. heterozygous
   d. true breeding
   e. F1 generation

7. Which of the following statements about the pedigree shown below is false?
   a. The mode of inheritance is autosomal recessive.
   b. The genotype of individual II-2 is aa.
   c. The genotype of individual II-3 is AA.
   d. The phenotype of individual III-3 is affected.
   e. The genotype of individual II-1 could be Aa or AA.

8. Which of the following genetic techniques is correctly matched with its use?
   a. karyotyping: used to identify DNA mutations
   b. gene testing: used to identify abnormalities in chromosome structure
   c. pedigree: used to calculate the probability of inheriting a genetic disorder
   d. gene therapy: used to replace a defective gene with a normal one
   e. karyotyping: used to determine the genotype of an individual

Answer the questions below.

9. Provide three reasons why pea plants were an excellent choice for Mendel’s research.

10. Describe the genotypes of the P generation commonly used in the series of genetic experiments to determine the inheritance pattern of a trait. Why is this important?

11. Given the phenotype of an individual, can you determine its genotype? Explain.

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

13. What is the expected phenotypic ratio in the offspring of a dihybrid cross between two heterozygotes?
14. List the gametes that could be produced from the following parents:
   a. AaBb
   b. AABbCc

15. How can you identify an autosomal dominant disease from a pedigree?

16. Sickle cell anemia is an autosomal recessive disorder that results in the formation of abnormally shaped red blood cells. Write the genotypes for the following individuals:
   a. a person with sickle cell anemia
   b. a person carrying the sickle cell allele
   c. a homozygous person with a normal phenotype

**Thinking and Investigation**

17. Describe an experimental approach to determine which of two alleles for a gene is recessive.

18. A black-haired true-breeding guinea pig is crossed with a white-haired true-breeding guinea pig. All of the offspring have black hair.
   a. Which hair colour is dominant?
   b. What are the genotypes and phenotypes of the parents?
   c. What are the genotypes and phenotypes of the offspring?

19. In humans, drooping eyelids are dominant to non-drooping eyelids.
   a. What are the two possible genotypes that a person with drooping eyelids might have?
   b. A man who is heterozygous for drooping eyelids and a woman with non-drooping eyelids have children. What are the expected genotypes and phenotypes of their children?
   c. A man with non-drooping eyelids and a woman with drooping eyelids have three children, all with non-drooping eyelids. How do you explain this outcome?

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

21. A yellow-haired rat is mated with a black-haired rat. Over time, the rats produce 45 black offspring and 52 yellow offspring. From these results, determine the probable genotypes of the parents and offspring. How could you determine the dominant allele?

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

23. The pedigree below traces the appearance of a cleft chin in a family. Is a cleft chin dominant or recessive? Explain your answer.

24. Osteogenesis imperfecta (OI), also known as brittle bone disease, results in extremely fragile bones that tend to break for no apparent reason. The following pedigree traces one form of OI in a family. Based on the pedigree, what inheritance pattern does OI display? Identify the phenotypes and genotypes of all of the individuals in this pedigree. If there is a genotype that cannot be determined, explain why.
Communication

25. Among some breeds of cattle, there is a trait called *polled*. Polled cattle do not develop horns. A farmer breeds the same two polled individuals several times and five calves are born. Three of these calves are polled, and two are not. Is the inheritance pattern for polling autosomal dominant or autosomal recessive? Explain using a Punnett square.

26. A child who does not have dimples or freckles is born to a man who has both and a woman who has neither.
   a. Draw a pedigree to illustrate the inheritance of dimples and freckles.
   b. Describe the inheritance pattern for both traits.
   c. Determine the genotypes and phenotypes of all three individuals.
   d. Determine the probability of these parents having another child who has freckles but no dimples.

27. Using a graphic organizer, distinguish between the types of genetic testing. Include what each test analyzes, and a disorder that can be diagnosed using the test.

28. Write an opinion paragraph on whether genetic testing for *BRCA1* and *BRCA2* mutations should be a standard medical test for all women that is covered by OHIP.

29. Research into gene therapy continues to be controversial. Do you believe the potential benefits outweigh the risks? Working in pairs, develop a debate for opposing sides of this issue.

30. **BIG IDEAS** Variability and diversity of living organisms result from the distribution of genetic materials during the process of meiosis. Choose one example of genetic diversity in plants or animals that was discussed in this chapter and that supports this statement. Illustrate the link between it and the processes of meiosis in a poster, pamphlet, or multimedia presentation.

31. **BIG IDEAS** Genetic and genomic research can have social and environmental implications. Certain populations and cultural groups interest genetic researchers because they have unique genetic characteristics. Identify, through research, an example of a genetic disorder that is studied in this manner. Describe the disorder and the study, including why that group was chosen. Prepare a presentation that also addresses social and ethical concerns about this type of research, such as who benefits the most from it and who should be allowed access to the genetic information of the people in the study.

32. Summarize your learning in this chapter using a graphic organizer. To help you, the Chapter 5 Summary lists the Key Terms and Key Concepts. Refer to Using Graphic Organizers in Appendix A to help you decide which graphic organizer to use.

Application

33. A genetic disorder in Scottish terriers is Von Willebrand’s disease, which is a blood-clotting disorder. Knowing what you know about inheritance, what questions should you ask a breeder to ensure that your new Scottish terrier puppy will not be affected by this disease?

34. In humans, albinism is recessive to normal pigmentation. Two of your friends have normal pigmentation. They have one child who is albino and are expecting a second child. Your friends tell you, “We knew that our chance of having one albino child was 1:4, so we are sure that our next baby will not be albino.” How would you respond? Using a specific example, explain to your friends whether their reasoning is correct.

35. People who are heterozygous for a recessive disorder do not express the trait, but they may pass it on to their children. Do we have a responsibility to inform our children of a recessive disorder they may have inherited from us? Why?

36. Research one example of a genetic disorder.
   a. Identify the gene and mutation that are associated with the disorder.
   b. Describe the test(s) for this disorder.
   c. What are important considerations for people who are considering getting tested for the disorder?
   d. What are potential options for people who test positive for the disorder?

37. Imagine that you are a genetic counsellor. A couple, Taku and Sara, come to see you. They are starting a family and want to know if their children are at risk for cystic fibrosis. Taku’s sister has cystic fibrosis. Taku and his parents do not.
   a. Draw a pedigree for the family. Indicate Taku and Sara on the pedigree.
   b. What questions should you ask in order to provide the best risk assessment? Explain.
   c. What further testing suggestions might you make?
   d. What are some concerns that this couple may have regarding testing and the results?
Select the letter of the best answer below.

1. **K/U** Which of the following describes the law of segregation?
   a. Each individual receives one copy of each gene from each parent.
   b. Each individual receives two copies of each gene from each parent.
   c. Each individual receives two copies of some genes from one parent, and two copies of the remainder of the genes from the other parent.
   d. Each gene segregates independently from all other genes.
   e. Each gene segregates independently from all other genes unless they are on the same chromosome.

2. **K/U** A gene exists in two different forms, A and a. Which type of gamete can a homozygous recessive individual produce?
   a. Aa
   b. A
   c. a
   d. A and a
   e. AA

3. **K/U** A cat with normal ears is crossed with a cat that has curled ears. All the kittens born from that cross have normal ears. Later, when these offspring are crossed with each other, the phenotypic ratio is 3:1 normal ears to curled ears. What conclusion can be made about the inheritance of curled ears?
   a. Curled ears are a result of crossing over.
   b. Curled ears are a dominant phenotype.
   c. Curled ears are a recessive phenotype.
   d. Curled ears are a hidden trait.
   e. More crosses need to be done to determine how the trait is inherited.

4. **T/I** An individual with the genotype AA is crossed with an individual with the genotype Aa, producing 120 offspring. The number of offspring with genotype AA will be close to
   a. 120
   b. 90
   c. 60
   d. 30
   e. 0

5. **K/U** If a black guinea pig (Bb) were crossed with a white guinea pig (bb), what would be the expected phenotypic ratio of the offspring?
   a. 0:1 black to white
   b. 1:0 black to white
   c. 1:1 black to white
   d. 1:3 black to white
   e. 3:1 black to white

6. **K/U** To determine if an individual displaying the dominant phenotype is heterozygous, the individual is crossed with an individual that is
   a. homozygous recessive for the trait
   b. homozygous dominant for the trait
   c. heterozygous for the trait
   d. of the same genotype as the individual in question
   e. of the opposite genotype as the individual in question

7. **K/U** In garden peas, purple flowers (P) are dominant to white (p) flowers, and tall plants (T) are dominant to short plants (t). If a purple tall plant (PpTt) is crossed with a white short plant (pptt), what is the expected phenotypic ratio of the offspring?
   a. 1:1:1:1 purple tall to purple short to white tall to white short
   b. 3:2 purple tall to purple short
   c. 9:3:3:1 purple tall to purple short to white tall to white short
   d. 1:1 purple tall to purple short
   e. all purple tall

8. **K/U** The allele for polled cattle is dominant to the allele for horned cattle. Occasionally, a horned calf appears in a herd of polled cattle. What is the most likely reason?
   a. All of the cattle are homozygous for the polled allele.
   b. Some of the cattle are heterozygous for the polled allele.
   c. Polled is the dominant allele.
   d. A mutation has occurred.
   e. Something unrelated to genetics has caused this result.

Use the following information to answer questions 9 and 10.

9. **K/U** How many affected males and females are in the pedigree?
   a. 1 male, 2 females
   b. 2 males, 1 female
   c. 3 males, 9 females
   d. 4 males, 5 females
   e. 5 males, 4 females
10. **A** Which of the following disorders could be represented by the pedigree?
   a. Down syndrome (chromosome 21 trisomy)
   b. Chronic myelogenous leukemia (translocation of chromosome 9 to chromosome 22)
   c. Cri du chat syndrome (deletion in chromosome 5)
   d. Tay-Sachs disease (autosomal recessive)
   e. Huntington disease (autosomal dominant)

*Use sentences and diagrams as appropriate to answer the questions below.*

11. **T/I** Distinguish between the following pairs of terms using a definition and an illustrative example.
   a. homozygous and heterozygous
   b. dominant and recessive

12. **A** A certain breed of cattle in England produces good meat when a particular gene is heterozygous. That same gene is lethal when it is homozygous recessive. How would a farmer produce the best meat without losing any cattle?

13. **T/I** In tomato plants, tall vines are dominant to short vines. A tomato plant heterozygous for vine length is crossed with a short-vined plant. What is the probability of producing a plant with tall vines?

14. **T/I** Purple flowers are dominant to white flowers, and tall plants are dominant to short plants. Outline an experiment to determine the genotype of a tall plant producing white flowers. Describe how the results can be used to identify the genotypes of the parent plants.

15. **T/I** In tomatoes, round shape is dominant to pear shape, and smooth skin is dominant to fuzzy skin. A farmer crossed two tomato plants, and obtained the results below. Determine the genotypes of the parents.

<table>
<thead>
<tr>
<th>Characteristics</th>
<th>Number of Offspring</th>
</tr>
</thead>
<tbody>
<tr>
<td>Round, smooth</td>
<td>213</td>
</tr>
<tr>
<td>Round, fuzzy</td>
<td>72</td>
</tr>
<tr>
<td>Pear-shaped, smooth</td>
<td>69</td>
</tr>
<tr>
<td>Pear-shaped, fuzzy</td>
<td>20</td>
</tr>
</tbody>
</table>

16. **C** Use your understanding of meiosis to illustrate the chromosome theory of inheritance.

17. **K/U** A pedigree is constructed over several generations. Why must information be collected for many generations?

*Use the following information to answer questions 18 and 19.*

A male affected by an inherited genetic disorder and an unaffected female have three children. The oldest child is an affected male, and the other two children are unaffected females.

18. **C** Create a pedigree from the information provided. Label the generations and the individuals.

19. **T/I** Is it possible to identify the pattern of inheritance from this information? If so, provide it. If not, explain why not.

20. **C** A male and a female are both heterozygous for a peaked hairline, which is the dominant phenotype. Construct a pedigree showing this mating, and the four potential offspring. **Hint:** Construct a Punnett square first.

21. **K/U** Cystic fibrosis is the most common fatal genetic disorder for young Canadians.
   a. Describe the genetic basis of this disorder and the genetic test used for its diagnosis.
   b. How is this disorder inherited within a family?
   c. What are potential concerns regarding social discrimination and ethical issues?

22. **A** If you just learned of a genetic disorder in your family, would you want to be tested? Why or why not?

23. **C** Huntington disease is often described as an autosomal dominant lethal disease. Do you agree or disagree with this statement? Explain your position.

24. **T/I** In addition to knowledge about inheritance patterns, what other skills do you think genetic counsellors should have? Explain why.

25. **A** “Gene therapy holds the promise of curing genetic disease, yet it remains controversial.” Discuss this statement using specific examples.