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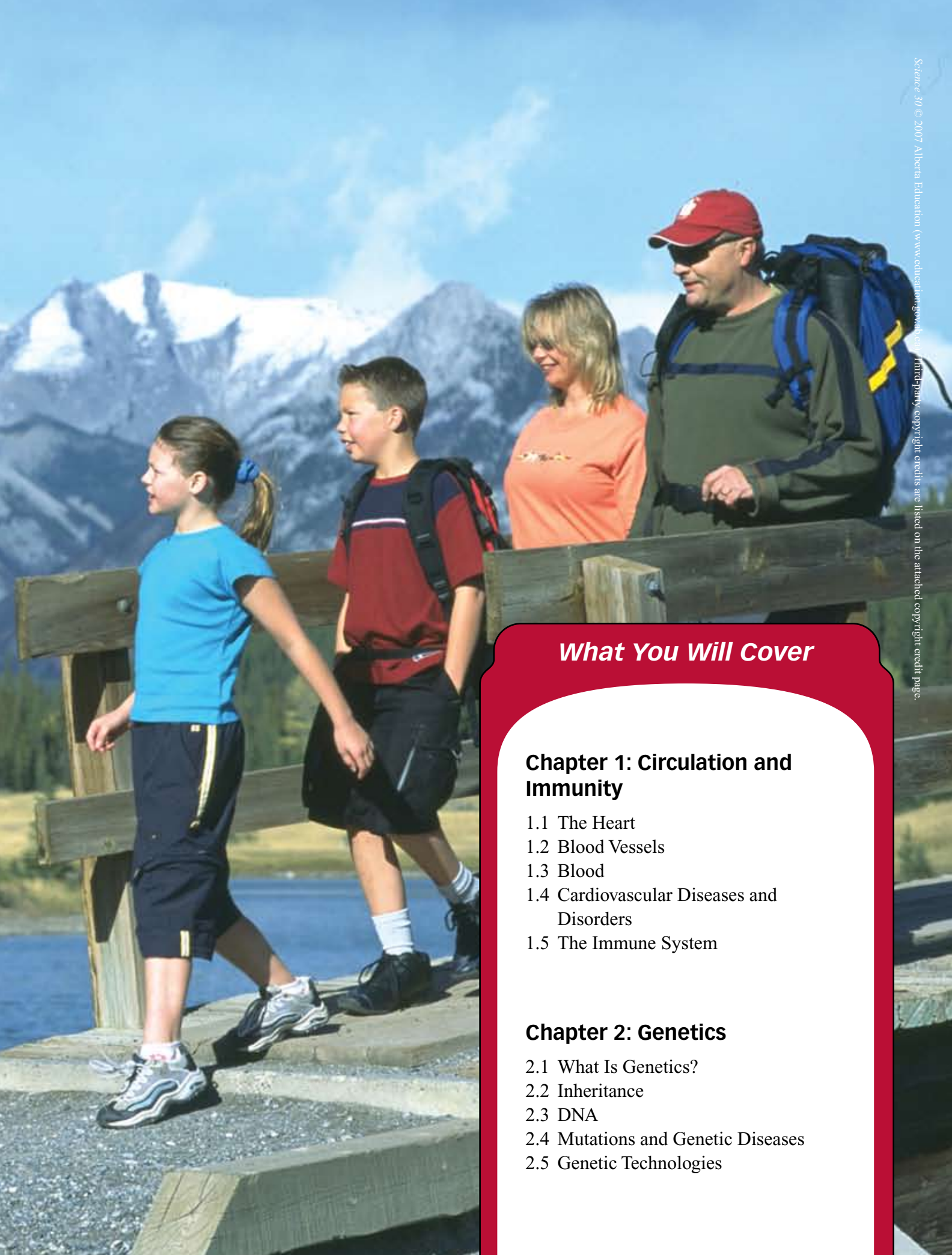
Unit A Maintaining Health

How would you spend a warm summer day in the mountains? For this family, hiking seemed like the best idea. Since the trail was not far from their campsite, they were able to start hiking by mid-morning.

The family in the photo enjoys these sorts of activities, and they look forward to many more years together of this kind of exploring. Their passion for hiking is just one of the many characteristics that they share: they all seem to enjoy high levels of fitness; they rarely get colds or the flu; and the children are frequently told that they strongly resemble each other and their parents. Which of these characteristics relate to lifestyle choices? Which ones are inherited? When you think about your own health and the characteristics that you share with your family members, which traits are due to the choices your family makes regarding diet and exercise, and which ones are inherited?

In this unit you will examine how the circulatory and immune systems work together to keep you healthy. You will study the major components of these two systems and consider factors that can affect how they function. In the second part of Unit A, you will look at the major principles of genetics and use these concepts to explain how some traits can be passed on from one generation to the next. Throughout the unit you will investigate and evaluate technologies used to explore, maintain, repair, and assist our bodies.



What You Will Cover

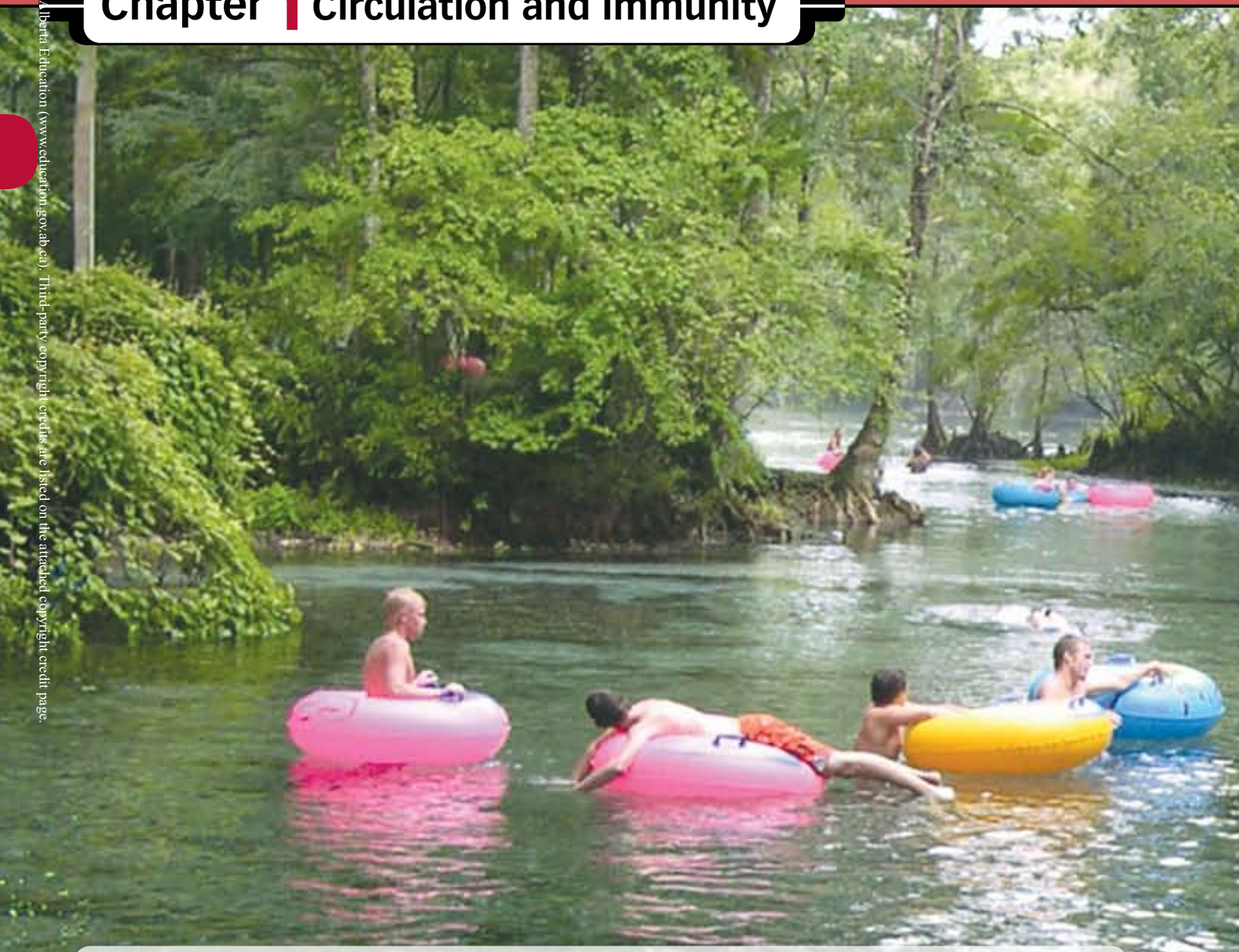
Chapter 1: Circulation and Immunity

1.1 The Heart
1.2 Blood Vessels
1.3 Blood
1.4 Cardiovascular Diseases and Disorders
1.5 The Immune System

Chapter 2: Genetics

2.1 What Is Genetics?
2.2 Inheritance
2.3 DNA
2.4 Mutations and Genetic Diseases
2.5 Genetic Technologies

Chapter 1 Circulation and Immunity



When was the last time you spent a hot summer's day on the water with your friends? Although kayaking requires some specialized equipment, tubing is wonderfully low tech—all you need is an inner tube and the right setting. Tubing down a river can be especially fun. Some businesses take advantage of places where a river naturally makes a C-shaped turn—this allows both the entry point and the exit point to be close together. In this way when a passenger reaches the exit point, it's a short walk uphill to start all over again. Places that rent inner tubes often employ lifeguards to make sure that the riders are safe. These businesses also employ security personnel to ensure that only paying customers use the facility's inner tubes. There is also a maintenance staff to replace worn-out tubes. Through local patterns of evaporation and precipitation, the water cycle provides the water pump for the river system.

The function and parts of the human circulatory system are comparable to tubing down the river. The circulatory system has a muscular pump that cycles blood. Some blood cells are shaped like the tubes so they can easily move through the bloodstream, but these cells transport dissolved gases instead of human riders. Like blood vessels, the river's banks direct the flow of fluid. Human blood has cells that act like the security guards, the lifeguards, and even the maintenance crews.

Try This Activity

Measuring Your Heart Rate Before and After Exercise

Your heart rate provides valuable information about your health. Immediately after exercising, athletes often compare their heart rate to their resting heart rate. This procedure monitors their level of fitness and the intensity of their training program.

CAUTION!

If you have a medical condition that prevents you from participating in physical education classes, you should not participate in the exercising part of this activity.

Purpose

You will measure your heart rate in three different situations: when you are resting; immediately after you have finished exercising; and at two-minute intervals after exercising.

Procedure

Using a watch or clock, count the number of beats in fifteen seconds and then multiply by four to get the beats per minute. It is best to measure your pulse two or three times and calculate an average to get the most accurate heart rate while you are resting. Record this number as your resting heartbeat in beats per minute (bpm).

step 1: Make sure you are seated and rested before beginning this activity. Locate your pulse or the pulse of a partner by using both your index finger and your middle finger. The pulse is most easily found by pressing these two fingers against the inside of your wrist or against the carotid artery, which runs up your neck on either side of your throat. Each beat of your pulse corresponds to a beat of your heart. Using your thumb may interfere with counting since the thumb contains its own pulsing artery.

step 2: Most members of your class will engage in four minutes of the **same** physical activity at a moderate level. Physical activities to choose from include jumping jacks, running on the spot, or, while seated, repeatedly lifting two textbooks from your shoulder to above your head. At the instant the activity ends, take your pulse. In beats per minute, record your pulse immediately after exercising.

step 3: Continue recording your pulse rate every minute for the next five minutes or until your pulse returns to its resting rate.

step 4: Record your average values for both your resting heart rate and your recovery time. Share this information with your teacher so that you and your classmates can calculate average values for your class.

Analysis

1. Compare your resting heart rate with the class average. Should a difference between heart rates alarm you?
2. List some factors that might contribute to the difference in resting heart rates among class members.
3. Describe how your heart rate changed during exercising, and relate how it was altered after you stopped.
4. How long did it take for your heart rate to return to its resting rate? Compare your time to recover with that of other people.
5. Explain why it is necessary for each class member to perform the same exercise for the same length of time.
6. Include in your health file your resting heart rate, your heart rate during exercise, and your recovery time.

As you learn about the circulatory system, immune system, and genetics, you will be collecting information about yourself similar to the information that a doctor might collect about you. Throughout Unit A you will compile this information into a health record for yourself. When you see the health file visual cue, add information to your file. In addition to recording valuable data about yourself, your health file will be a valuable study guide.

1.1 The Heart

Transportation Systems

The human body is made up of trillions of cells closely packed together. These cells are similar to the closely packed houses that make up a city. Each house's inhabitants generate wastes that must be regularly removed, and each house requires a constant supply of water and energy (such as electricity and natural gas). The houses are often far from the source of the needed supply or the waste disposal site. Like houses, cells generate wastes and require constant supplies. An efficient network for transporting materials is required to keep both cells and houses functioning properly. Blood vessels in the human body function very much like highways, roads, and pipes that serve cities and towns. Notice that no home in the photo is far from a road. In your body, no cell is more than two or three cells away from a blood vessel. Like roadways, there are one-way blood vessels, major and minor blood highways, and even the occasional traffic jam as blood vessels break or clog.

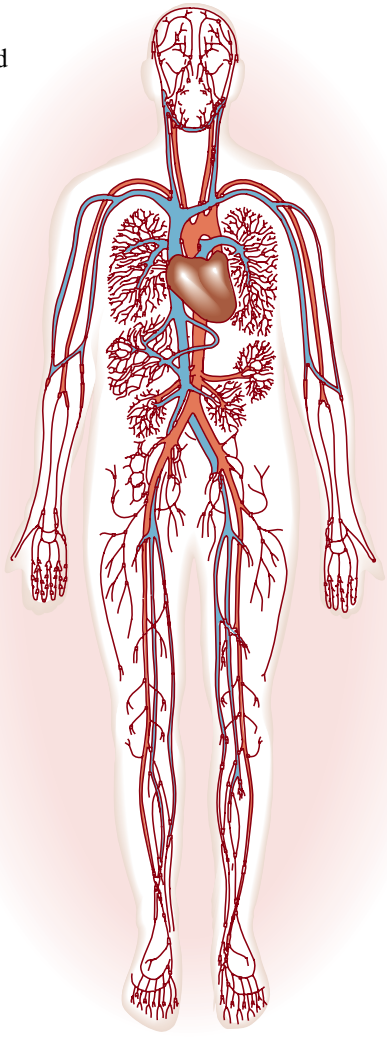
The Body's Internal Transportation System

Microscopic organisms and even some larger invertebrates do not need to have an extensive internal transportation system. This is because their cells are in direct contact with the environment, and gases and materials can move to each of the organism's cells through simple diffusion. Similarly, as cities grow larger, a greater number of roadways and services and more complicated networks for transportation are required. The larger and more complex the organism, the greater the need for a more extensive internal transportation network that effectively transports materials to all specialized cells. This internal transportation network is called the **circulatory system** or the **cardiovascular system**.

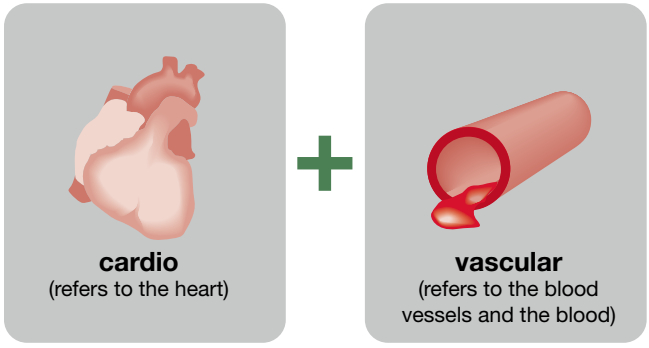
circulatory system or cardiovascular system: the system consisting of the heart, blood vessels, and blood that circulates through the body

The human circulatory system performs four key functions. It

- transports and delivers oxygen and nutrients (e.g., minerals, vitamins, and glucose) to the body's cells in exchange for carbon dioxide and wastes
- transports and delivers chemical messengers—such as hormones—throughout the body
- distributes body heat
- defends against disease



The heart, blood vessels, and blood are the circulatory system's major components. Many people use the term *cardiovascular system* because this name includes the key parts of



By the end of Chapter 1, you will be able to describe the structure and function of the circulatory system and its major parts and examine how the circulatory system facilitates interactions between the human body's blood cells and the external environment. You will also investigate substances that harm the circulatory system and study disorders of the system.

Ideas About the Heart

Some historical ideas about how the heart and circulatory system work may seem strange to some. The ancient Egyptians believed that a person's emotions, wisdom, and personality originated in the heart rather than in the brain. When someone died they believed that the dog-headed god, Anubis, weighed that person's heart to determine his or her fate in the afterlife. Even today, people still use this Egyptian idea of the heart causing emotions by using metaphors like "suffering from a broken heart," "stealing someone's heart," or "speaking from the heart."

In the second century CE, a Greek physician named Galen became very influential as a personal physician to the Roman emperor. He was very interested in observing the functioning of biological systems. Since studying human dissections was not considered acceptable, many of Galen's ideas were based on his studies of animal dissections. This led him to develop misconceptions about human anatomy, including that the heart was split into two chambers, that food was turned into blood by the liver and then used up by the body, and that blood sloshed back and forth like the ocean's tides. Galen also believed that the heart sucked blood in from the veins rather than acting like a pump. Galen's ideas were widely accepted and his misguided teaching influenced beliefs that lasted for an incredible 1500 years!

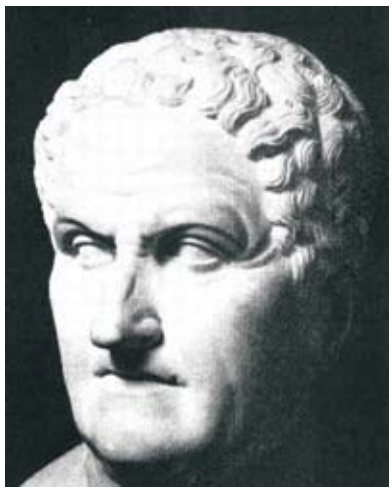
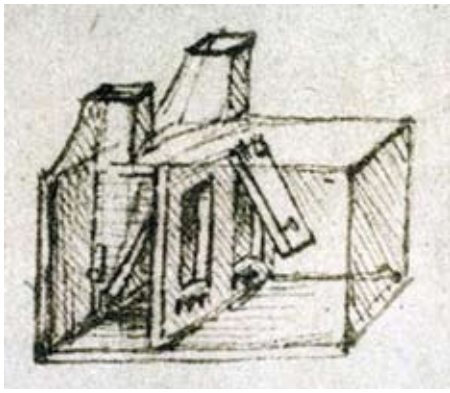
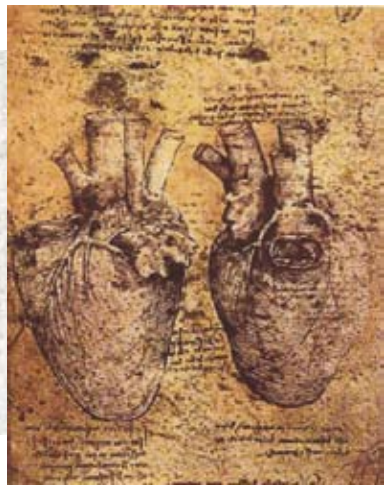


Figure A1.1: Galen

Leonardo da Vinci





In the late fifteenth century, Leonardo da Vinci, the famous Renaissance artist, inventor, and scientist began examining many human cadavers and made accurate and detailed drawings of the heart and circulatory system. Da Vinci made careful records of his observations, often comparing the human body to a machine. One of his drawings compares the human heart, with its chambers, to a furnace. Experimentation and investigation on human cadavers was discouraged at the time and some of da Vinci's findings contradicted the beliefs and teachings of that era. To keep his work secret and to prevent other people from stealing his ideas, he used his own special shorthand mirror image writing. Perhaps if circumstances had permitted Leonardo da Vinci to share and publish his work, a more complete understanding of the heart and its functions would have been available to Renaissance-era physicians.

William Harvey

In the 1600s the physician William Harvey began to seriously question the teachings of Galen. Findings based on Harvey's studies of human anatomy and dissections of human cadavers disagreed with Galen's well-established theories. Harvey found that there were valves in both the heart and the veins that kept blood moving in one direction—not sloshing like ocean tides as Galen believed. By using simple mathematical calculations, Harvey took the volume of blood that the heart could hold and multiplied that volume by the number of times the heart beat per minute. This calculation resulted in a value far greater than the amount of fluid the body could hold. Harvey concluded that the heart must be re-pumping the same blood. This experiment contradicted Galen's idea that the liver was turning food into new blood to be used up by the body. Because Harvey was not able to explain how blood got from the arteries to the veins, his theory was not immediately accepted.

Figure A1.2: William Harvey

A few years after Harvey's death, Marcello Malpighi used a microscope to discover the tiny hair-like capillaries that connect arteries to veins. This confirmed Harvey's theory that a small volume of blood was constantly circulated to all parts of the body and that the heart, arteries, and veins were connected in a circulatory system.

cardiac output: the volume of blood pumped by the heart in one minute, which is equal to the product of stroke volume and heart rate

At the centre of Harvey's work were simple calculations relating to the volume of blood and the heartbeat. Today, similar calculations can reveal amazing information about the effectiveness of the heart as a pump. The heart of a typical adult human male pumps out 70 mL of blood per beat. This is called stroke volume. The stroke volume for a typical adult human female is about 60 mL per beat. The average resting heart rate for men and women is around 72 beats per minute. So, if you know the volume of blood pumped in each beat and the number of beats that occur in a minute, you can determine the volume of blood pumped by the heart in one minute. This value is called the **cardiac output**.

Example Problem 1.1

A typical human male has a stroke volume of 70 mL per beat and a resting heart rate of 72 beats per minute.

a. Calculate the cardiac output. Express your answer in litres per minute.

b. Calculate the volume of blood that would be pumped in one day based upon the cardiac output.

Solution

a. stroke volume = 70 mL/beat

$$= 70 \text{ mL/beat} \times \frac{1 \text{ L}}{1000 \text{ mL}}$$

$$= 0.070 \text{ L/beat}$$

heart rate = 72 beats/minute

cardiac output = ?

$$\text{cardiac ouput} = (\text{stroke volume}) \times (\text{heart rate})$$

$$= \frac{(0.070 \text{ L})}{\text{beat}} \times \frac{72 \left(\text{beats}\right)}{\text{min}}$$

$$= 5.04 \text{ L/min}$$

$$= 5.0 \text{ L/min}$$

The cardiac output is 5.0 L/min.

Note: It is often best to handle the conversion of units when listing the data. The final answer is rounded to two significant digits since the given values are expressed to two significant digits.

b. volume pumped in one day $= \frac{(5.04 \text{ L})}{\text{min}} \times \frac{(60 \text{ min})}{1 \text{ h}} \times \frac{(24 \text{ h})}{1 \text{ d}} = \frac{7257.6 \text{ L}}{\text{d}} = 7.3 \times 10^3 \text{ L/d}$

Note: The unrounded value from a. is used in the follow-up calculation in part b. Since the original given values were expressed to two significant digits, the final answer should be rounded to two significant digits. In this case, scientific notation is required, unless the answer is expressed in kL.

Practice

1. Copy and complete this table. In the middle column, summarize each person's different historical theories about the heart and circulatory system. Each of the three listed individuals contributed at the time to the understanding of the heart and circulatory system. In the right column, describe the limitations of each theory or contribution.

HEART THEORIES

Person	Theory About the Heart	Limitations of the Theory
Galen		
Leonardo da Vinci		
William Harvey		

2. A female Science 30 student has a resting heart rate of 68 beats per minute. Determine her cardiac output in litres per minute.

Include your cardiac output in your health file.

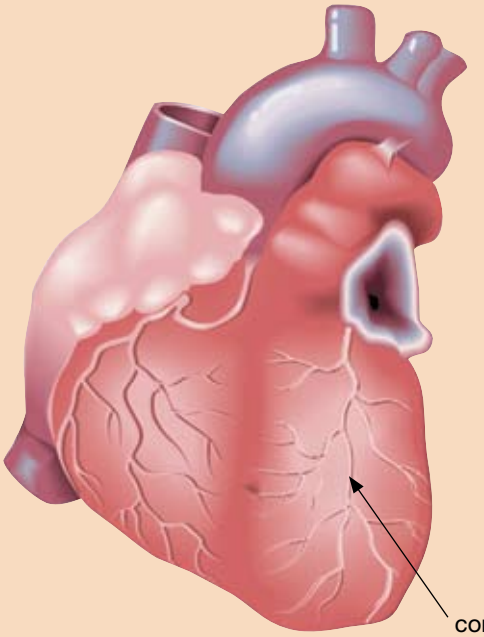
3. Refer to your answer from question 2. How much blood would this student's heart pump in one year if it maintained the resting heart rate?

4. The average human has approximately 5 L of blood. How long does it take the heart to pump this volume?

5. If a male raised his heart rate to 180 beats per minute through intensive exercise, such as running on a treadmill, how much blood would his heart pump per minute? Assume that the stroke volume remains at 70 mL per beat.

6. A rain barrel holds approximately 213 L of water. This same volume could fill more than 100, 2-L pop bottles.

- **a.** Determine your resting heart rate using the techniques from "Try This Activity: Measuring Your Heart Rate Before and After Exercise." Use this value to calculate your cardiac output.
- **b.** If you had a pump working at the same rate as your cardiac output, how long would it take to fill a 213-L barrel?
- **c.** Suppose your heart rate doubled its resting value because you were exercising. How long would it take your pumping heart to fill the same barrel under these circumstances? Assume the stroke volume remains constant for all parts of this problem.



The Heart: An Amazing Pump

Clench your hand into a fist. The size of your closed hand corresponds approximately to the size of your heart. Now squeeze your hand and relax it. Imagine doing that action about every second. That would add up to over 80 000 times per day and 2.5 billion times in an average lifetime! This squeezing—called contracting—and relaxing is exactly what your heart does every day.

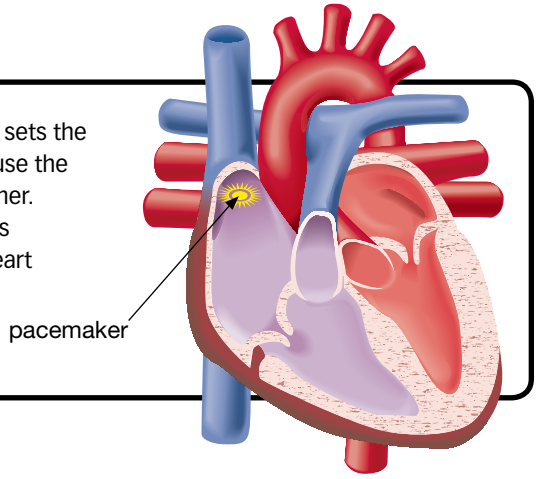
Like any other muscle that is contracting, the heart needs a constant supply of oxygen and other nutrients. Since the entire body depends upon the heart, the first organ that the heart supplies with oxygen-rich blood is itself. The blood vessels that supply the heart are called the **coronary arteries**.

coronary arteries: the vessels that supply the heart muscle with oxygen-rich blood

DID YOU KNOW?

The pacemaker is a small region of specialized muscle tissue that sets the tempo of the heartbeat. The pacemaker generates electrical signals that cause the muscle fibres in the heart to simultaneously contract in a co-ordinated manner. If one muscle fibre of a heart chamber is stimulated to contract, all the fibres of that chamber contract in unison. This en masse contraction makes the heart muscle unique.

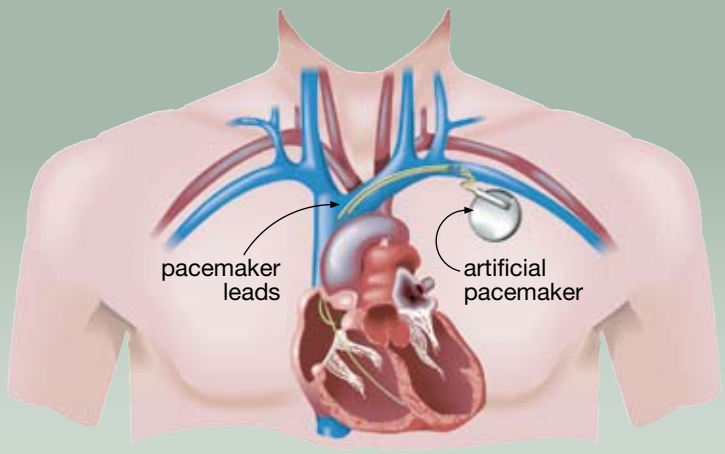
If the heart's pacemaker cells are unable to regulate a steady heartbeat, then an artificial pacemaker can be surgically implanted.



Science Links

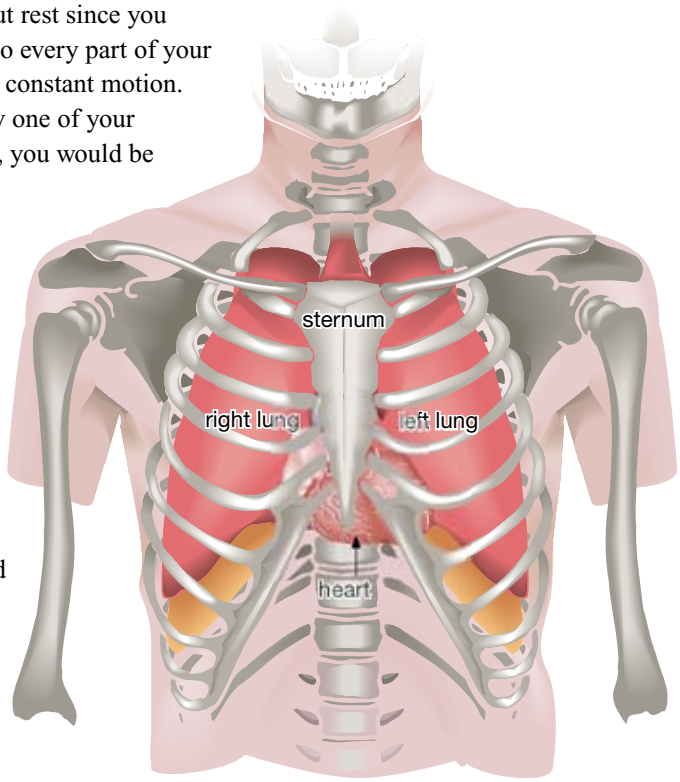
An artificial pacemaker is a small battery-operated machine that sends electrical signals to the heart through tiny wires. This little computer "listens" to the heart and supplements its normal rhythm. Adjustments to the pacemaker can be made without further surgery by using radio-wave signals. These signals are sent through the skin and other tissues to the artificial pacemaker from a control wand outside of the body.

In Unit C you'll learn more about radio waves as well as devices that both produce and transmit electrical energy.



Your heart is a muscular pump that has been beating without rest since you were a developing embryo, and it will continue to push blood to every part of your body until the end of your life. All the blood in your body is in constant motion. With each powerful heartbeat, life-giving blood is sent to every one of your approximately 60 trillion cells. Without the work of your heart, you would be dead in a matter of minutes.

When you imagine a heart you might think of the familiar shape that children learn to draw in preschool, but the shape of the human heart is really more like an upside-down pear with four open spaces, or chambers, inside. Since the heart is a muscle, its look and texture is like that of the red meat you would see in a raw steak. Contrary to the popular belief that the heart is found on the left side of the chest, the heart is actually located almost in the centre of the chest where it is protected by the hard sternum (or breastbone). During a medical checkup, the doctor listens on the left side with a stethoscope because the heart is pointed slightly to the left, and sounds produced by parts of the heart are easier to hear there.



Anatomy of the Heart

Study the labelled diagram of the human heart in Figure A1.3. Note that the areas containing oxygen-rich blood are shaded red and the areas containing oxygen-poor blood are blue.

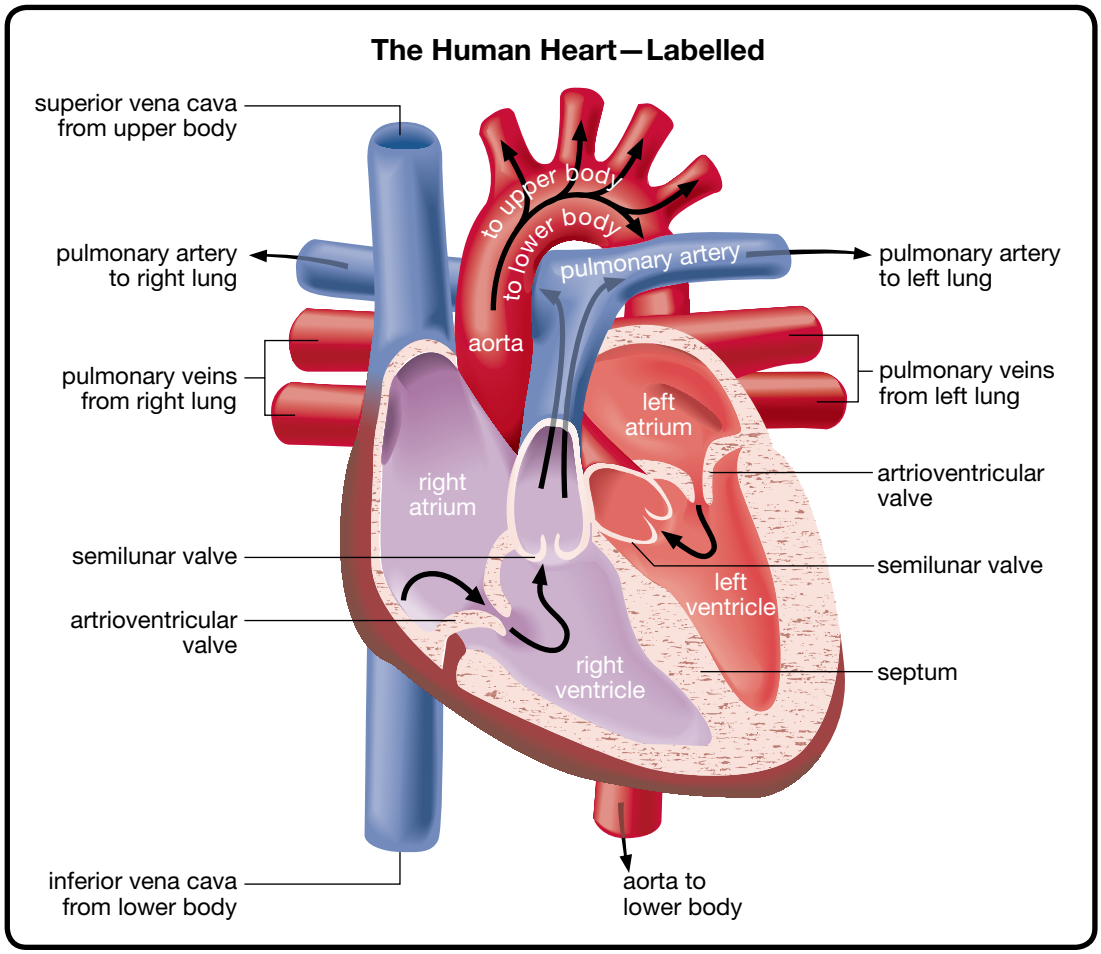


Figure A1.3

Do you notice that the human heart in Figure A1.3 seems to have the left and right mixed up? That's because heart diagrams are labelled from the point of view of the person who has the heart. If this heart was in the person who was facing you, this is how this person would label left and right.

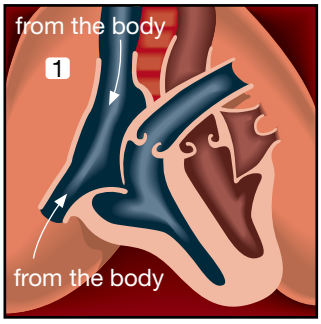
You probably also noticed that the heart is unevenly split into four chambers. The right and left sides of the organ are partitioned by a thick wall called the **septum**. The smaller top two chambers are the left **atrium** and right atrium, together called atria. The bottom two pointed chambers are the left **ventricle** and right ventricle. The left ventricle is slightly bigger in size because its job is to pump oxygen-rich blood to most of the body. The four chambers are divided by **heart valves** that ensure blood will travel in only one direction through the heart. The valves between the atria and the ventricles are held in place by string-like tendons that act like the ropes on a drawbridge. These tendons help ensure the proper alignment of the valves when they are closed.

- **septum:** a thick wall of muscle that divides the left and right sides of the heart
- **atrium:** the smaller upper chamber that receives blood returning to the heart
- **ventricle:** the larger v-shaped bottom chamber that pumps blood from the heart
- **heart valves:** thin flaps of tissue in the heart that open and close to ensure the proper direction for blood flow

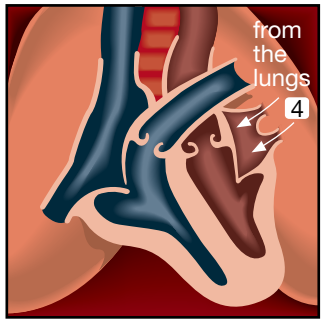
Blood Flow Through the Heart

Since blood circulates constantly through the body, you could begin to trace the flow of blood at any point. In Figure A1.4, blood's path is traced in a step-by-step manner starting with the place where blood first enters the heart on its way back from the body.

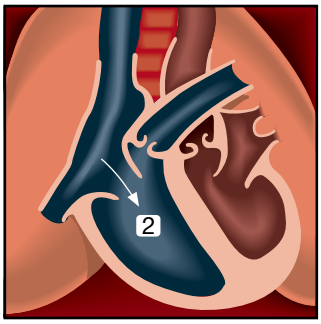
Figure A1.4



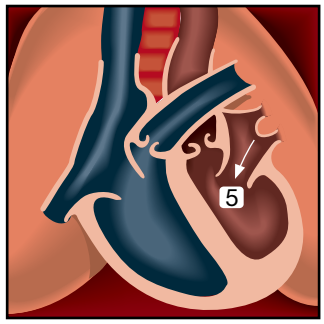
1. The vena cavae are large veins that collect oxygen-poor and carbon dioxide-rich blood from the upper body and lower body. The blood enters the heart and collects in the right atrium from both the superior and inferior vena cavae.



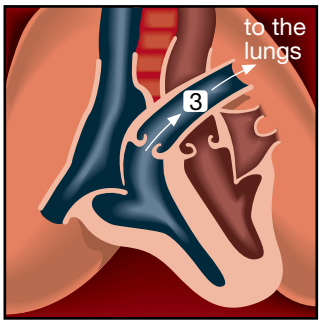
4. The oxygen-rich blood is then sent back toward the heart through the pulmonary veins to the left atrium.



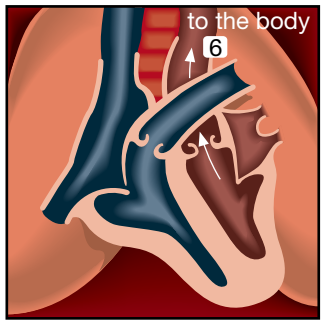
2. The contraction of the right atrium forces blood into the right ventricle.



5. The contraction of the left atrium forces the blood into the left ventricle, the heart's most muscular chamber.



3. The right ventricle contracts and forces the blood to flow out of the heart into the pulmonary arteries. The pulmonary arteries move the blood away from the heart toward the right and left lungs. At the lungs, the blood undergoes gas exchange by receiving oxygen and releasing carbon dioxide, a waste product of cell activity.



6. The contraction of the left ventricle forces the blood into the aorta. The aorta is the largest artery in the body and through its many branches, directs oxygen-rich blood to the entire body.

Practice

7. Obtain the handout "The Human Heart" from the Science 30 Textbook CD. Attempt to answer the following questions without referring to Figure A1.3. After you have completed as much as you can, use the diagrams in the textbook to complete and/or correct your work.

   a. Add a label for each of the areas identified on this diagram.

   b. Add red shading to the areas that deal with oxygenated blood and blue shading to the areas that deal with deoxygenated blood.

   c. Add arrows to indicate the direction of blood flow through each of the chambers and major blood vessels.

   d. Add the numbers *1* through *6* to outline the sequence for the path of a blood cell as it travels through each of the heart chambers. Begin with number 1 representing the place where blood first enters the heart on its way back from the body.

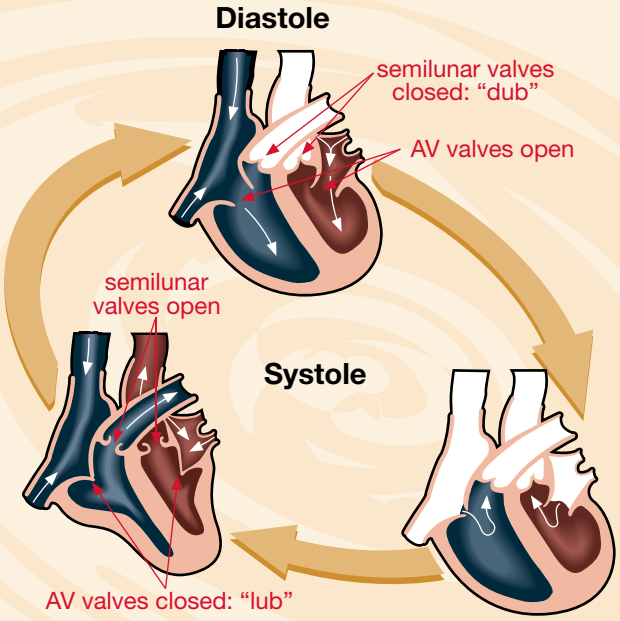
Heartbeat

You have so far traced the step-by-step flow of blood through the heart, but the action of the heart does not work in steps. Blood does not move through one chamber while the other chambers lay empty waiting for their turn to move the blood on; instead, the two sides of the heart fill at the same time and act together like parallel pumps. Once filled with blood, both atria contract at the same time, followed by the simultaneous contraction of both ventricles. Before the contraction of the ventricles occurs, they are relaxed and the valves between the atria and ventricles are open. This allows blood to flow in and fill the ventricular chambers. This relaxation part of the cycle is called the **diastole**. In the first step of a two-step contraction, the atria contract together to push the blood down into the ventricles. In the second step, the two ventricles contract to force the blood out of the heart. This two-part contraction of the heart cycle is called the **systole**. The "lub-dub" heart sound that a doctor listens for through a stethoscope is due to the heart valves functioning during diastole and systole.

One complete contraction (systole) and one complete relaxation (diastole) combine to make a heartbeat—one cycle of the heart's activity.

diastole: the phase of the heart's cycle where a chamber of the heart, either an atrium or a ventricle, relaxes and fills with blood

systole: the phase of the heart's cycle when the ventricles contract to eject blood from within the chamber



The atria and ventricles relax, (atrial and ventricular diastole) filling with blood.

The artrioventricular valves open due to the lower pressure of the blood within the ventricles.

Blood outside the heart is under higher pressure, so the semilunar valves are forced to close. A "dub" sound is made.

When the atria contract—called atrial systole—the blood is forced from the atria into the ventricles.

The artrioventricular valves remain open.

The semilunar valves remain closed.

The two ventricles contract (ventricular systole) to force blood out of the heart.

The high pressure forces the artrioventricular valves to close. A "lub" sound is made.

The semilunar valves are forced open by the high-pressure blood leaving the ventricles.

Utilizing Technology

The Animated Heart

Purpose

You will have an opportunity to observe the systole and diastole phases of the heart's cycle by using the applet "The Human Heart." This applet is located on the Science 30 Textbook CD.

Science Skills

✓ Analyzing and Interpreting

Procedure

step 1: Select "Human Heart." Then scroll to "Heart Parts." Practise naming each part of the heart, and then move the cursor over each part to confirm your prediction.

step 2: Select the applet part called "Animated Heart." Carefully watch this animation. Focus on the action of the valves. As the animation plays, add a soundtrack by saying "lub" and "dub" at the correct times. Adjust the heart rate and observe the differences in your spoken sound track.

step 3: Select the part called "Narrated Tour." Locate the position of your own heart as you listen to the description.

Analysis

Computer animations can demonstrate complex processes in ways that are clear and easy to understand. However, compromises are made in terms of which details are included and which ones are omitted. Watch the computer animation again as you answer the following questions.

1. Which details of the systole part of the heart's cycle are included? Which are omitted?
2. Which details of the diastole part of the heart's cycle are included? Which are omitted?
3. If a person has a heart rate of 72 beats/min, then one heartbeat lasts 0.83 s. In other words, the entire heart cycle of diastole, atrial systole, and ventricular systole occurs in just 0.83 s. On average the diastole lasts for 0.4 s and the ventricular systole lasts for 0.3 s, which only leaves about 0.1 seconds for the atrial systole.

Use this information to suggest an explanation for the trends you identified in your answers to questions 1 and 2.

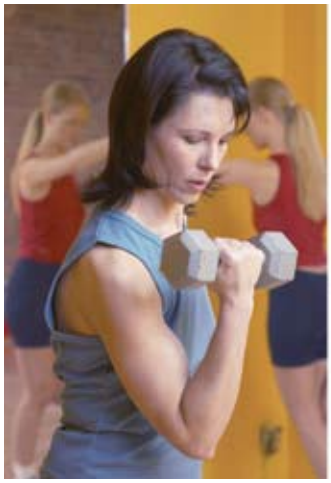
DID YOU KNOW?

When a doctor listens to a patient's heart with a stethoscope, sometimes swishing or whooshing sounds—called *heart murmurs*—are heard in addition to the standard "lub-dub" of the heartbeat. Heart murmurs result from the turbulent flow of blood through the heart—this is why they can sound like water rushing through the end of a garden hose.

Pediatricians classify most heart murmurs they hear as *innocent*, because they are not associated with a heart disease or abnormality. Most children will have a heart murmur at some time, but these innocent murmurs usually disappear by the time they become adults.

The doctor may decide that the characteristics of a particular heart murmur require additional testing to determine if there is an underlying problem with blood flow through the heart. In these cases, the murmur is usually due to the abnormal functioning of a heart valve. A valve may not be closing tightly—it may be too narrow or too stiff. A treatment plan is then designed to address the specific condition.

Factors Affecting Heart Rate



Many factors can affect your heart rate. From your own experiences you know that emotions, such as fear or excitement, quickly increase the heart rate. At one time, you have probably been so scared that it felt like your heart was going to jump out of your chest. Changes in external temperature also can cause your heart rate to change. For example, if you sit in a hot tub, the external temperature of your body increases greatly and the heart must work harder to pump blood around in an attempt to dissipate body heat through the skin.

If training with weights can make a muscle like your bicep larger, can other forms of exercise make your heart larger?

Exercise that improves your heart's ability to provide working muscles with oxygen is called cardiovascular training, or aerobic exercise. Examples include swimming, running, or cycling. In each case, the exercise is done non-stop at a moderate rate for at least 20 minutes. These activities are commonly called cardio workouts because they increase the demand of the body's muscles for oxygen. Cardio workouts, therefore, cause the heart to increase the volume of blood it pumps every minute, elevating the heart rate above its resting value.

The effect of a lifestyle that includes cardiovascular exercise is that the heart and surrounding blood vessels do not become larger; but, instead, these tissues improve in their stretching ability. If the heart muscles are more elastic they have a greater capacity to expand, thus increasing the amount of blood pumped during each heartbeat. These improvements to elasticity translate to an increased stroke volume both when the heart is put under peak demand and when it is resting. This is why people who engage in a lot of cardiovascular exercise develop hearts that need to beat less often to circulate the same amount of blood. A stronger heart is not a larger heart, but is instead a more elastic one. Athletes tend to have a lower-than-average resting heart rate—often only 45 to 50 beats per minute.

Utilizing Technology

Heart Rate Monitoring

Athletes often record their heart rates immediately after waking and record this data for an extended period of time. This technique gives them more accurate resting-heart-rate data because there are probably fewer external variables first thing in the morning—such as diet, caffeine, or exercise—that could affect the heart rate. Increases in the morning resting heart rate could indicate the onset of illness or a lack of recovery from overtraining.

Science Skills

- ✓ Performing and Recording
- ✓ Analyzing and Interpreting
- ✓ Communication and Teamwork

Purpose

For two weeks you will record your resting heart rate and the minutes spent in physical activity every day.

Procedure

step 1: For two weeks you will record your resting heart rate before you get out of bed, and you will also note the approximate minutes spent in physical activity for each day. Physical activity could include walking, participation in sports, dancing, or movement associated with chores or a job such as carrying or lifting.

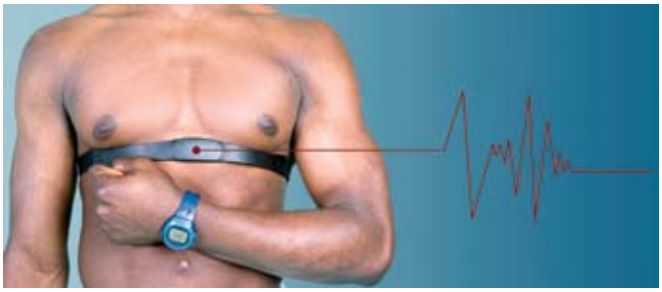
step 2: Organize your data using a spreadsheet. Use the spreadsheet to create graphs that summarize your results.

Analysis

1. Did you notice any significant changes within the resting-heart-rate data? Were these changes related to the onset of an illness or to a sudden change in the level of physical activity?
2. Compare your findings with those of other students. Is there evidence to support the idea that individuals who regularly participate in cardiovascular exercise tend to have a lower resting heart rate? Why is this question difficult to answer?

Target Heart Rate for Exercise

A chest-strap heart-rate monitor is often used to help ensure that the wearer is exercising at a desired level. Once your heart exceeds approximately 85% of its maximum heart rate, your body burns less fat and produces more lactic acid, which causes muscle soreness. One way to estimate your maximum heart rate is to subtract your age from 220.



It is important to realize that this method of determining your maximum heart rate is just a guideline. Some medications, especially those related to the heart, require a lower maximum heart rate be used than the one provided by this guideline. If you have a chronic medical condition or have any doubts about whether a medication you are taking affects your maximum heart rate, contact your physician to determine your maximum heart rate.

Target Heart Rates

Personal Health Goal	Percentage of Maximum Heart Rate
maintain fitness level	50 to 60%
increase fat burning or weight loss	60 to 70%
increase cardiovascular endurance	70 to 80%

The maximum heart rate is an important value because it helps provide a guide for goals that you may have for either maintaining or improving your health. A common mistake that people make is that they begin a new exercise program with activities that cause their heart rate to be too high. The best approach is to pace yourself by beginning with activities that will only push you to about 50% of your maximum rate. This is especially important if you have had an inactive lifestyle. You can then gradually increase the intensity of your workouts over the first months of your program. It is always a good idea to consult with your physician if you are just starting a new exercise program or if you have questions about health or fitness.

Practice

8. Determine your maximum heart rate by subtracting your age from 220.
9. Use your answer from question 8 to complete the following table.

Your Target Heart Rate

Personal Health Goal	Heart Rate
maintain fitness level	
increase fat burning or weight loss	
increase cardiovascular endurance	

10. Describe how your answers to question 9 will change as you get older.

Add your maximum heart rate and your target heart rate for different health goals to your health file.

DID YOU KNOW?

The most effective cardiovascular fitness programs involve activities that are done for at least twenty minutes four or five times a week at moderate activity levels. If you make walking your primary means of transportation, you can build a fitness routine into your day without having to join either a gym or a health club. As is the case with any fitness programs, the best approach is to build regular walking into your weekly routine, to gradually increase the intensity, and to include warm-up and cool-down stretches.

Investigation

Dissecting a Mammal's Heart

Purpose

You will identify the main parts of a dissected heart, and you will trace the path of oxygenated and deoxygenated blood through the heart. You will choose one of two possible pathways for this activity: one that uses instruments to dissect the heart of a mammal; and the other that involves a virtual dissection.

Science Skills

- ✓ Performing and Recording
- ✓ Analyzing and Interpreting

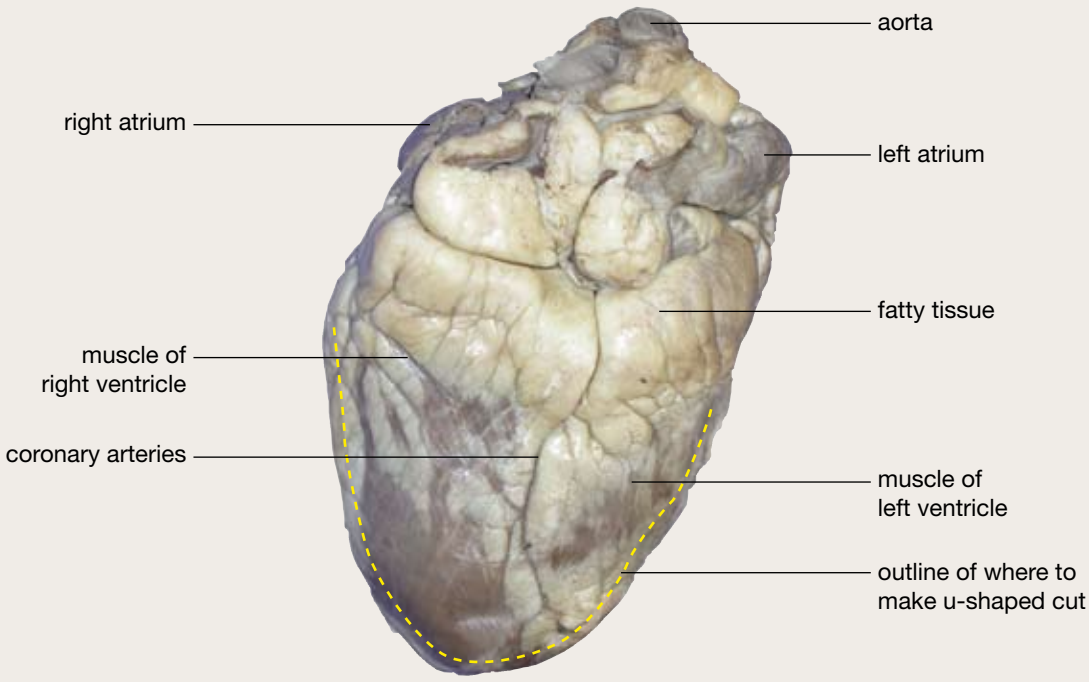
Part A: Using Dissecting Instruments and a Mammalian Heart

Materials

- heart of a mammal (pig, sheep, or cow)
- set of dissecting instruments—scalpel, scissors, probe, pins
- 2 pieces of yarn 40-cm long, one red and one blue
- handouts "Labels for the Parts of the Heart" and "The Human Heart—Labelled" from the Science 30 Textbook CD
- dissecting tray
- latex or vinyl gloves
- goggles
- apron
- digital camera

Procedure

step 1: Place the heart in front of you with the largest blood vessel—the aorta—at the top of the heart and facing the bottom of the dissecting tray. Notice the presence of a diagonal line of blood vessels, going from the upper left to the lower right on the outside of the heart. These are the coronary arteries that supply the heart itself with blood. These vessels are often surrounded by some fatty tissue.



Confirm that you have the heart oriented properly by feeling each half of the heart on either side of the coronary artery with your hand. The heart's left side should feel thicker and more muscular than the heart's right side. **Remember:** The heart's left side is on your right as you look at the heart.

step 2: Begin by making a u-shaped cut around the sides of the heart to make a "flip-top" heart. Carefully hold the scalpel parallel to the table top as you cut. Be sure not to completely cut the heart into two separate sections. You should end up with a "flip-top" heart. This allows the interior of the heart to be observed and also to be put back together for later in this activity.

step 3: Lift the upper side of the heart away to reveal its inner chambers. Identify the side of the heart with the thicker-walled chambers. This is the heart's left side. Orientate the heart so the heart's left side is on your right.

step 4: Using your finger or a dissecting probe, locate where the blood enters the right atrium (vena cava). Blue yarn can be used to simulate the pathway in which deoxygenated blood flows. Thread a piece of blue yarn through the vena cava into the right atrium and then through the atrioventricular valve into the right ventricle.

step 5: Use your finger or probe around to find out where the blood must exit the right ventricle through the semilunar valve into the pulmonary artery. Remember that the blood cannot go back through the one-way valve or across the septum, which forms a barrier between the heart's right and left sides. Thread the blue yarn from the right ventricle to the pulmonary artery.

step 6: Tie a piece of red yarn to the blue yarn to simulate that the blood has become oxygenated in the lungs. Use the red string to trace the pathway that oxygenated blood flows through the pulmonary veins, to the heart's left side, and out through the aorta.

step 7: Obtain the handout "Labels for the Parts of the Heart" from the Science 30 Textbook CD. Cut out each of the labels and use dissection pins to attach the labels to the corresponding parts of the heart. If you are uncertain, use the "The Human Heart—Labelled" handout to help you identify the major parts of your dissected heart.

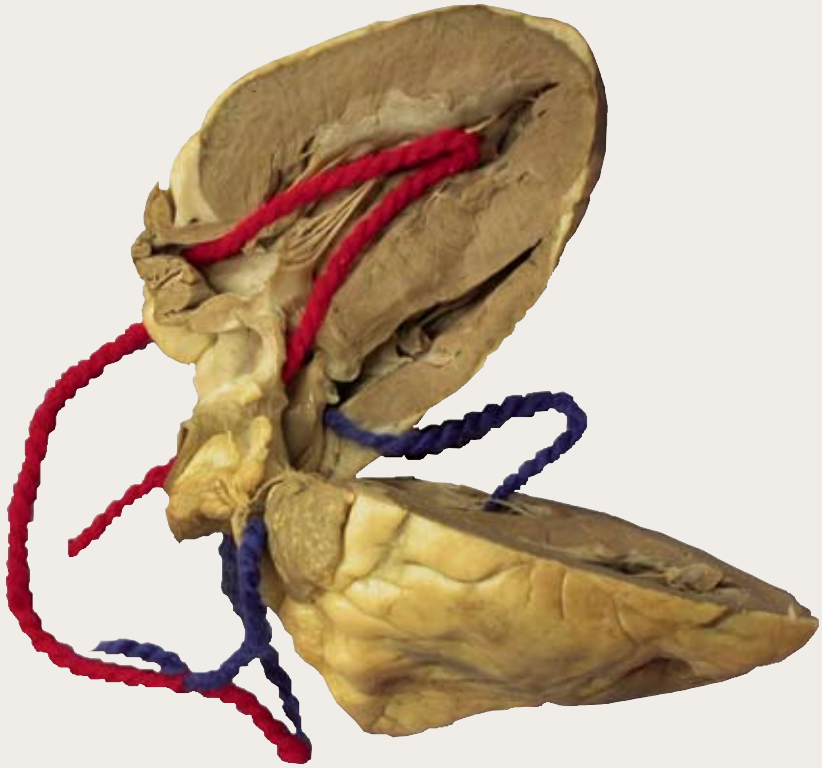
step 8: Have your teacher check your labelled heart.

step 9: Take a digital photograph of your dissected heart, complete with all the labels pinned in place.

Part B: Using a Computer Applet

Procedure

Locate the virtual version of "Dissecting a Mammal's Heart" on the Science 30 Textbook CD. Follow the directions on the applet as you complete your virtual dissection.



1.1 Summary

Beliefs about the heart and the circulatory system have changed over time. William Harvey was the first person to prove that blood circulated around the body in a closed system of vessels. The pump that drives the circulatory system is the heart. The output of blood from the heart depends on how many times the heart contracts and how much blood it moves with each contraction. The atria contract simultaneously, followed by the simultaneous contraction of the two ventricles. This two-part contraction creates a "lub-dub" sound due to the functioning of the heart's valves.

The heart rate is affected by emotion, temperature, exercise, fitness level, sleep, hormones, chemicals, drugs, and alcohol. By monitoring the heart rate during exercise programs, appropriate levels of exertion can be ensured. Heart rate is a key indicator of cardiovascular fitness.

1.1 Questions

Knowledge

1. Beginning with the vena cava, indicate the order of the following structures of the cardiovascular system through which blood flows: left atrium, right ventricle, lungs, body, right atrium, left ventricle, aorta.

2. Refer to Figure A1.5. Match the numbered structures on the heart to the part of the heart that

- **a.** receives oxygenated blood from the lungs
- **b.** sends oxygenated blood to the body
- **c.** prevents the backflow of blood in the heart
- **d.** separates the right and left halves of the heart
- **e.** collects deoxygenated blood from the body

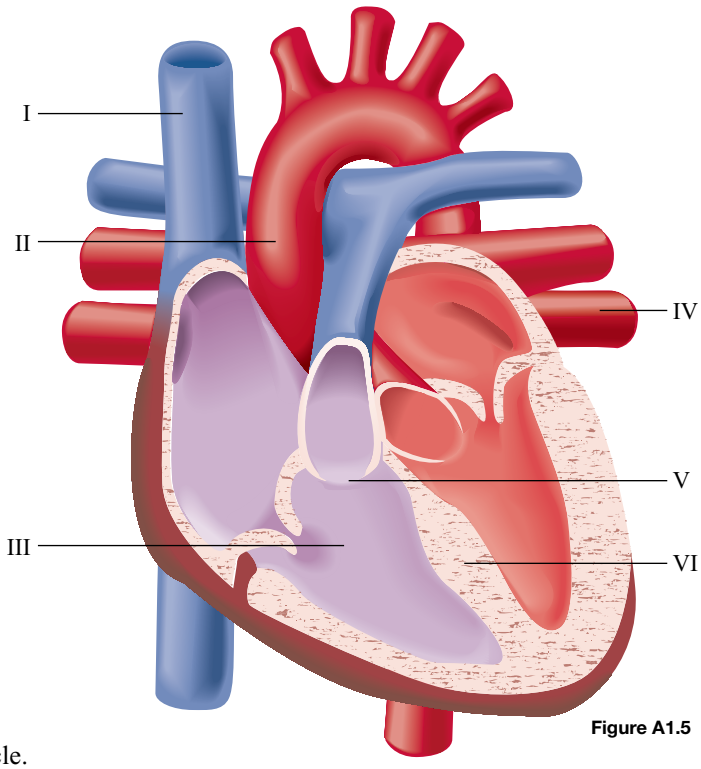


Figure A1.5

Applying Concepts

3. **a.** If an Olympic athlete has an increased stroke volume of 100 mL, calculate his cardiac output at rest (50 bpm), with light exercise (115 bpm), and with high-intensity exercise (180 bpm). Assume the stroke volume remains constant.

b. Explain why you expect the athlete to have a lower resting heart rate than a person with an inactive lifestyle.

4. Why are the walls of the heart's right ventricle thinner than the walls of the heart's left ventricle?

5. Compare the systole and diastole portions of the heart cycle.

6. Describe the purposes of a chest strap or other type of heart-rate monitor when a person is exercising.

1.2 Blood Vessels

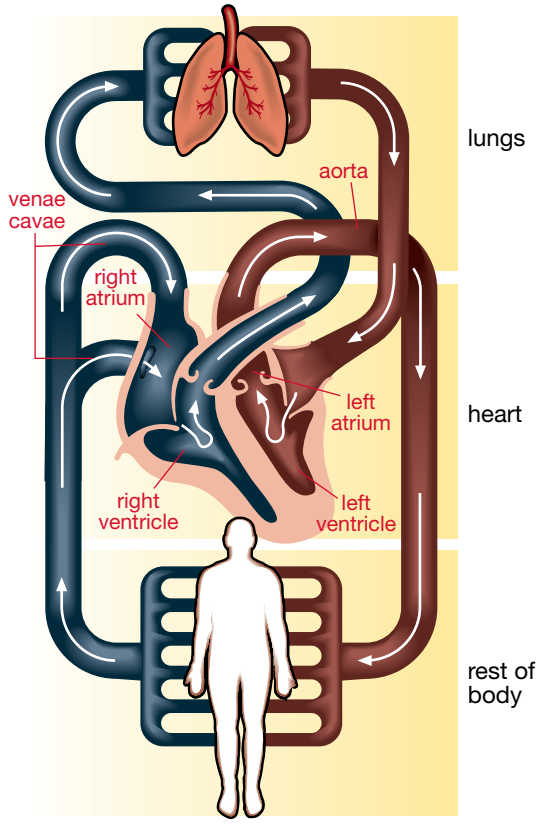
Figure A1.6: Glacial meltwater flows into the Bow River, which supplies about one million people with drinking water in southern Alberta.

The water that most Albertans drink begins as melt water high in the Rocky Mountains. Rivers then carry this water to communities throughout the province where it is pumped to water purification facilities and reservoirs before moving to people's homes. Each dwelling requires two piping systems: one to bring in the clean water and another to remove the waste water.

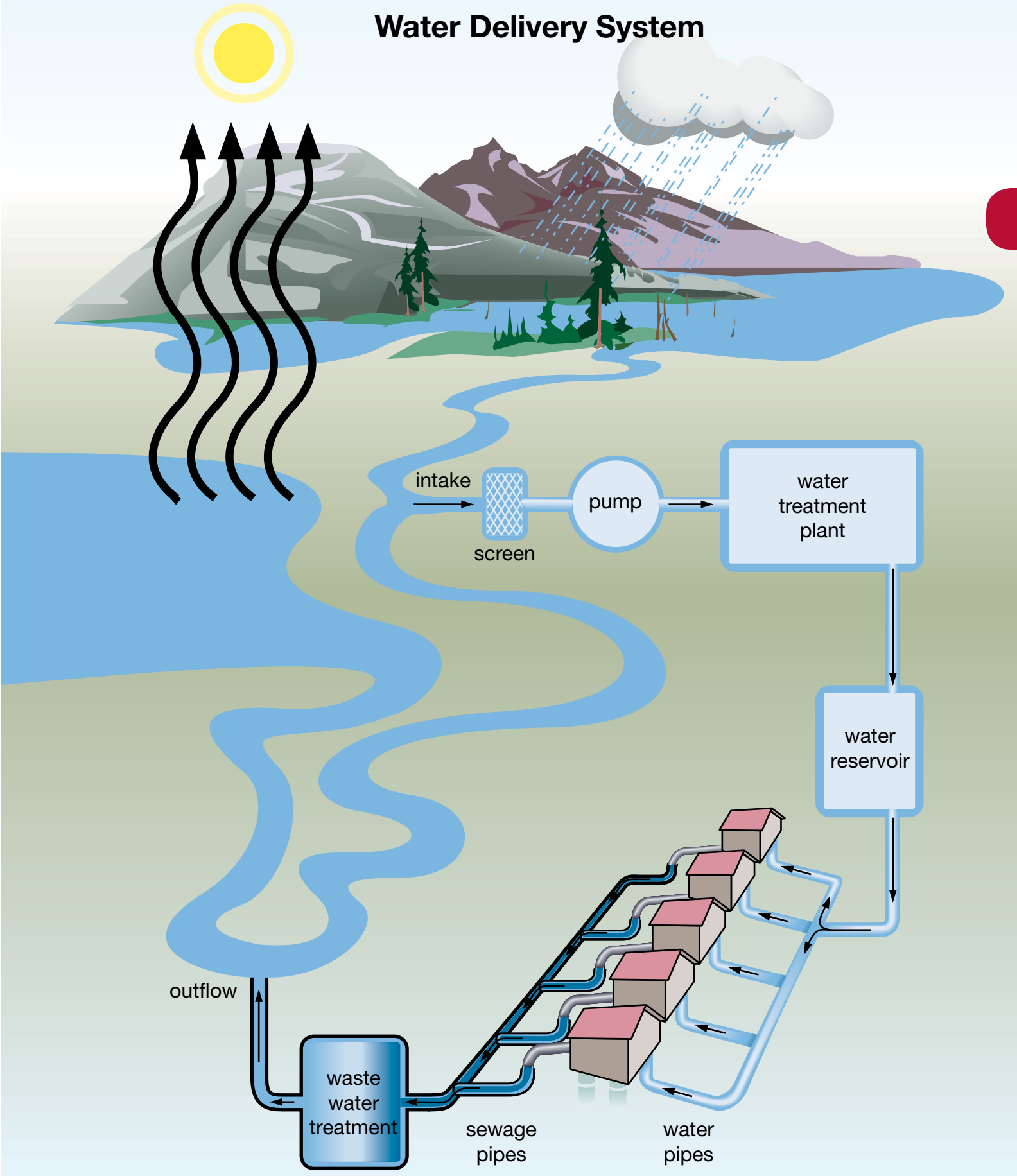
Because both systems transport fluids, the circulatory system of the human body can be compared to the water delivery system in a city or town. The cells in people's bodies have similar needs to the residents of a home. Every cell in your body, from the cells that make up the pumping muscles of the heart to the faraway cells in the tips of your toes, must be constantly supplied with blood. The sewage waste water collected at each home must be quickly removed and wastes must not be allowed to mix with the clean water coming from the pumping station, so wastes are transported in separate pipes. The blood that leaves the heart through the aorta is rich in oxygen and nutrients. This blood must be pumped in separate vessels from the blood that is full of carbon dioxide and wastes.

Just as cities and towns must have a fast and efficient way of transporting clean water to residents and of removing waste water, the body's circulatory system must also work to quickly and efficiently transport blood to and from the cells.

Human Circulatory System

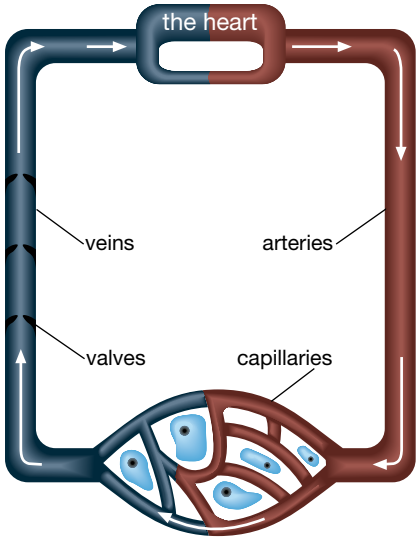


Water Delivery System



The Pathway of Blood

As you saw in Lesson 1.1, the basic components of the human circulatory system are the heart, which pumps the blood; the blood vessels that transport the blood to all parts of the body; and the blood.



Three types of blood vessels transport the blood. A blood vessel that carries blood away from the heart is called an **artery**, while a blood vessel that transports blood toward the heart is called a **vein**. Both arteries and veins branch into smaller vessels to effectively reach every part of the body. A **capillary** is a microscopic tube that connects the smallest branch of an artery to the tiniest branch of a vein. Capillaries are thin-walled porous vessels that allow materials, such as gases and fluids, to be exchanged with the body's cells. Every living cell in the body must be close to a capillary to remain alive and functioning.

To keep the facts straight about arteries and veins, many students use this memory device:

Arteries carry blood **A**way from the heart.

Major Arteries and Veins

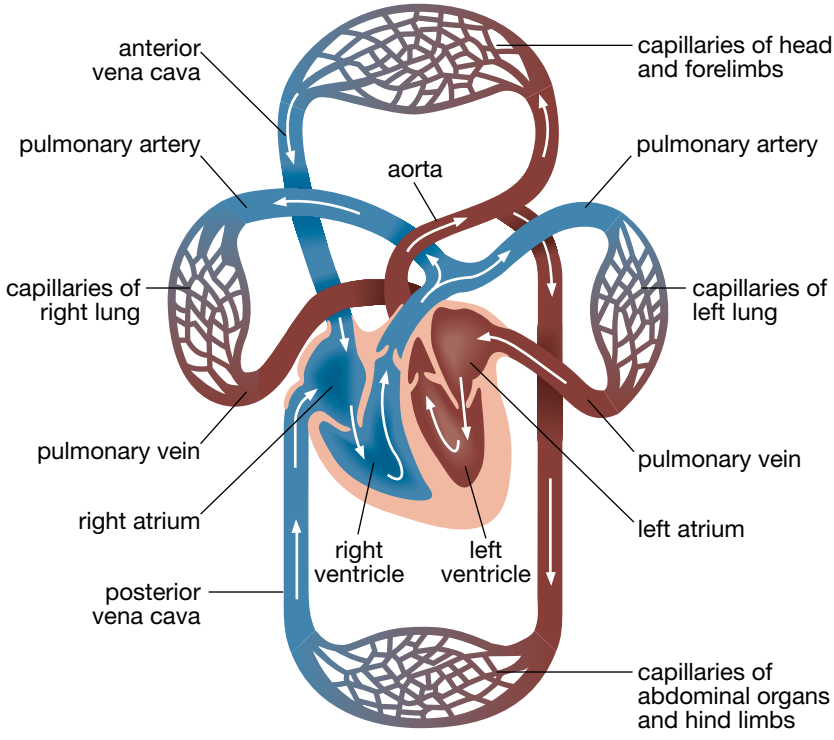
The deoxygenated blood arrives back to the heart from the body's major veins, which flow into the **venae cavae**, the largest veins in the body. The blood from each vena cava is carried to the heart's right side where it is pumped to the lungs through the **pulmonary arteries**. Within the capillaries of the lungs, the blood exchanges carbon dioxide for oxygen. The oxygen-rich blood then returns to the heart's left side through the **pulmonary veins**. The word *pulmonary* means "having to do with the lungs" because it comes from *pulmo*, the Latin word for lungs. This is why the movement of blood into and out of the lungs is called pulmonary circulation.

Except for the pulmonary vein, blood that flows through the veins is low in oxygen. Whereas oxygen-rich blood is red in colour, oxygen-poor blood is a darker shade of red. In diagrams, the difference in blood colour is emphasized by using completely different colours—red for oxygen-rich blood and blue for oxygen-poor blood.

Oxygen-rich blood leaves the heart by travelling through the largest artery in the body, known as the **aorta**. Recall that the first branches of the aorta are the coronary arteries, which supply oxygen and nutrients to the heart muscle itself. These arteries appear on the surface of the right and left ventricles. The aorta divides further into other arteries that carry blood to the major organs and body tissues.

- **artery:** a thick-walled blood vessel that carries blood away from the heart
- **vein:** a thin-walled blood vessel with valves that carries blood toward the heart
- **capillary:** a tiny blood vessel that connects the smallest branch of an artery to the smallest branch of a vein
- **vena cavae:** the largest veins in the body that carry oxygen-poor blood to the heart
- **pulmonary artery:** the large blood vessel that carries oxygen-poor blood from the heart's right ventricle to the lungs
- **pulmonary vein:** the large blood vessel that carries oxygenated blood from the lungs to the heart's left atrium
- **aorta:** the largest artery in the body; carries oxygen-rich blood from the left ventricle of the heart

Major Arteries and Veins



Practice

11. The body's circulatory system can be compared to a community's water system.

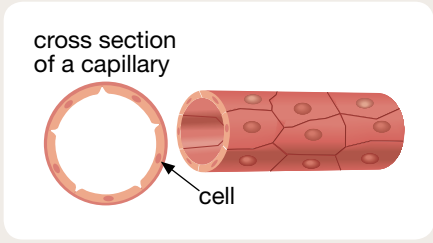
- **a.** Identify the parts of the body's circulatory system that correspond to the community's water pipes, sewage pipes, pump, and water.
- **b.** Describe at least two limitations of the comparison between a community's water system and the body's circulatory system.

12. Identify the major artery or vein that best matches each description.

- **a.** carries oxygen-poor blood from the heart to the lungs
- **b.** the body's largest artery
- **c.** carries oxygen-rich blood from the aorta to nourish heart tissues
- **d.** carries oxygen-rich blood to the heart
- **e.** carries oxygen-poor blood to the heart from the body's tissues

13. It is a common misconception that arteries always carry oxygen-rich blood and veins always carry oxygen-poor blood. Explain why this concept is not true in the case of pulmonary circulation.

14. A capillary is a microscopic structure. Its walls are comprised of only one layer of very thin cells.



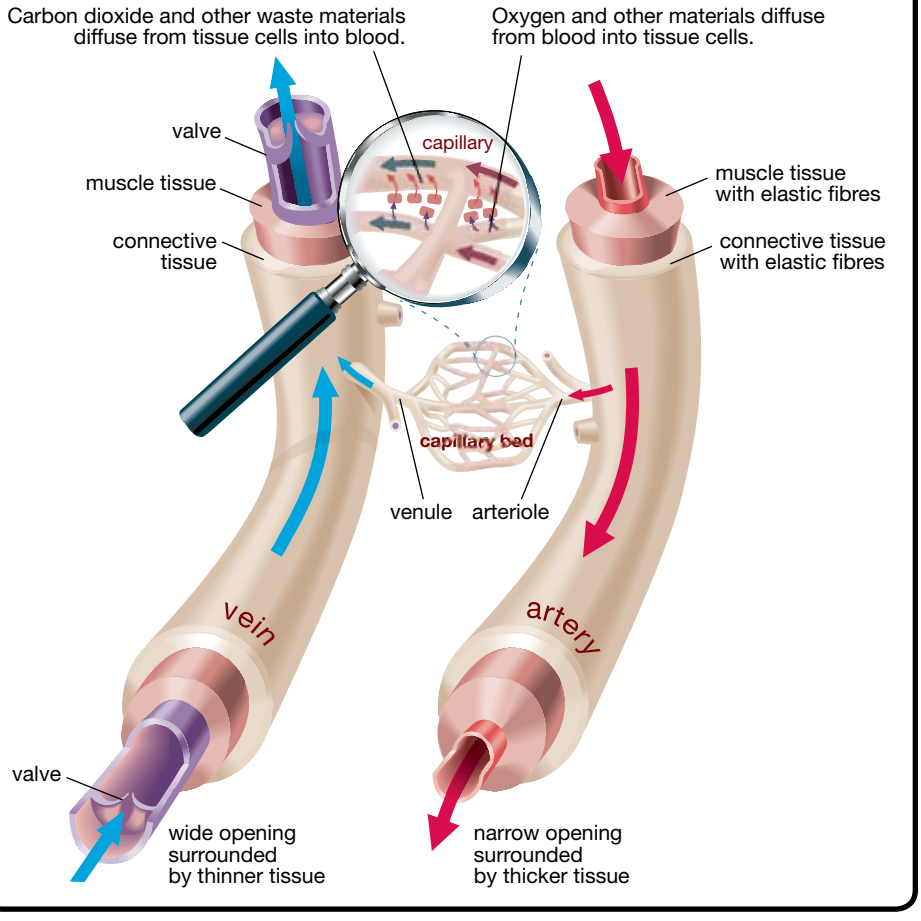
Explain why it is important for the walls of capillaries to be thin.

The Specialization of Blood Vessels

The blood vessels of the circulatory system are specialized for their specific functions. Arteries have thick elastic walls to withstand the pressure of the pumping heart. Except in the case of the pulmonary artery, the blood that flows in the arteries is oxygen-rich. This oxygenated blood is bright red in colour, and arteries other than the pulmonary artery are usually coloured red in circulatory system diagrams. As the arteries get farther away from the heart and aorta, they branch out and get smaller in diameter and lower in pressure. These smaller branched arteries are called **arterioles**.

arteriole: a small artery that joins a larger artery to a capillary

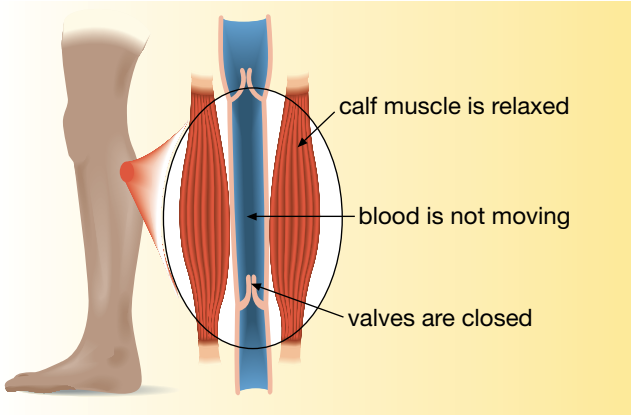
Arteries, Veins, and Capillaries

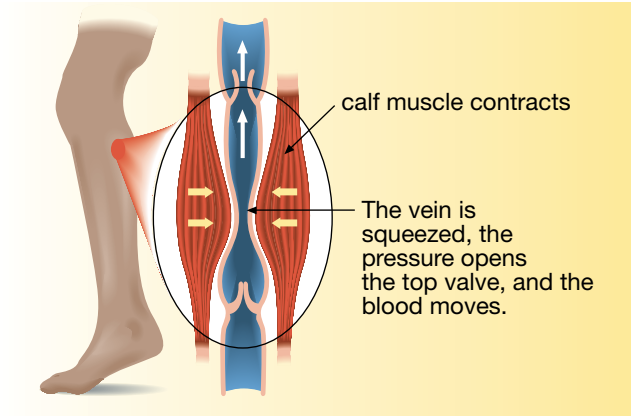


Arterioles are attached to the very thin-walled capillaries. The capillary is the place where needed nutrients, like oxygen and glucose, are exchanged for wastes—like carbon dioxide. The capillary walls are only one-cell thick because they need to be thin enough for the exchange of gases to take place by diffusion. Capillaries exist in a **capillary bed**, which is a web of capillaries surrounding the cells of body tissues. There are thousands of kilometres of capillaries in a human body. If you lined up all of the blood vessels end to end, they would wrap around Earth's equator at least four times!

- **capillary bed:** a network of capillaries in a particular area or organ of the body
- **venule:** a small vein that joins a larger vein to a capillary
- **varicose vein:** an enlarged, twisted vein near the surface of the skin resulting from poorly functioning valves

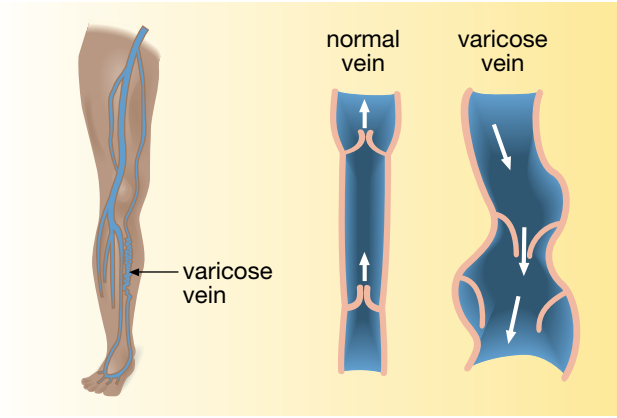
After the blood—depleted of oxygen and nutrients—leaves the capillary, it flows into the branches of veins called **venules**. The blood in venules and the larger veins has a much lower pressure than the blood pressure in an artery, so the walls of veins do not need to be as thick and elastic as the walls of arteries. The low-pressure blood has to get back to the heart against the pull of gravity. This is accomplished with the help of one-way valves in veins that prevent a backflow of blood, and also by the action of contracting body muscles.





Varicose Veins

As a person moves, the moving muscles push on blood in the veins while one-way venous valves prevent a backflow of blood and direct the blood back toward the heart. If the veins become stretched and the valves are damaged, blood in the veins pools and the veins become raised in a condition called **varicose veins**. People who spend much of their day standing have a greater tendency to develop varicose veins.



Practice

15. A blood cell travels through different blood vessels as it passes through the circulatory system after leaving the heart. The blood vessels involved include the following terms: capillary, vein, venule, artery, and arteriole.

Read each of the following descriptions and match each blood vessel term with a description.

- **a.** Large one-way valves in this vessel help direct blood back to the heart.
- **b.** These vessels are so small that blood cells must pass in single file.
- **c.** Capillaries converge into this vessel before entering a vein.
- **d.** This vessel is the pathway for oxygen-rich blood to enter capillaries.
- **e.** This vessel has thick walls with elastic fibres.

16. Consider the numbered list of blood vessels you used in question 15. Beginning with oxygen-rich blood that leaves the heart, place these terms in the order in which they are encountered by a blood cell.

17. Explain why circulatory problems often occur with people who are bedridden or with inactive people who seldom use their muscles.

18. Why do varicose veins most often occur in the lower legs?

19. Why should people who spend much of their workday standing up ensure that they elevate their feet at the end of the day?

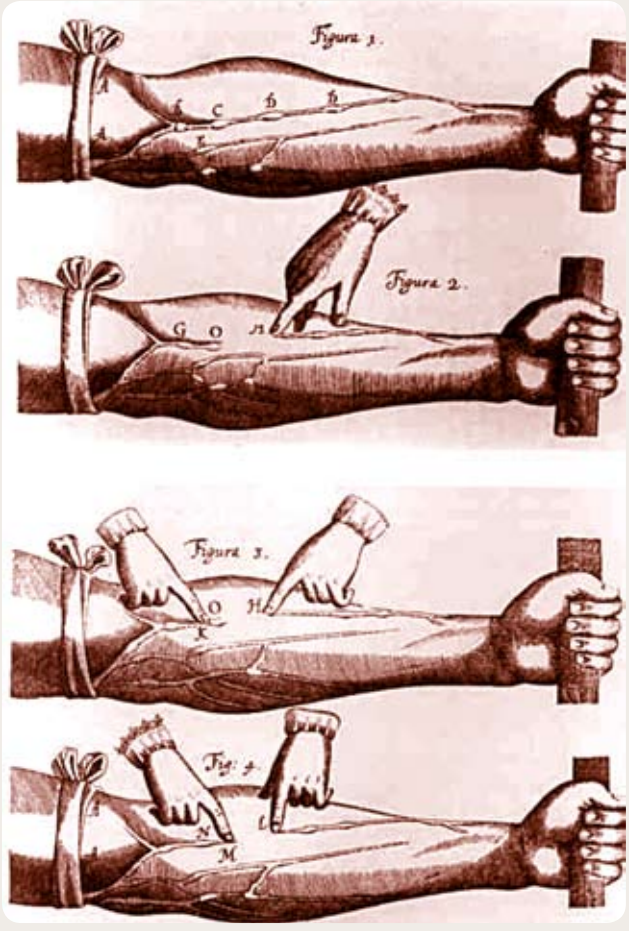
Try This Activity

William Harvey's Experiment

Science Skills

- ✓ Performing and Recording
- ✓ Analyzing and Interpreting

British scientist William Harvey helped prove that blood circulates in a closed system of blood vessels rather than swishing back and forth like tides, as was previously believed. Part of his investigation looked at the role of valves of the veins.



Purpose

You will observe the action of the valves working within the veins in the back of your hand.

Procedure

step 1: Lay your hand flat on a table so that you can see the veins on the top of your hand. Note that these photographs show the procedure using veins on your left hand.

step 2: Describe the appearance of the veins. Do the veins branch out or are they straight? Do the veins bulge more in certain areas? Record your observations.

step 3: Locate a straight section of a prominent vein. Firmly place the middle finger of your other hand on the end of this straight section of veins closest to your fingers.

step 4: While continuing to push down with your middle finger, take the index finger of your other hand and push down close to the middle finger on the same vein.

step 5: Continue to apply pressure with both fingers, and slide your index finger toward your wrist until you reach the end of the vein's straight section.

step 6: While continuing to apply pressure with your middle finger, release your index finger.

step 7: Carefully observe the straight section of the vein that used to be between the two fingers. Does blood flow back into the vein? Is there observable evidence of a valve's presence?

step 8: Repeat the process described in steps 3 to 7 with the following modifications:

- Place the index finger at the end of the straight section of vein closest to the wrist.

- Then place the middle finger next to the index finger. Slide the middle finger toward the fingers, away from the wrist.
- Release the middle finger and see if the blood flows back into the veins.

Observations

1. Describe your observations from steps 2, 7, and 8.

Analysis

2. State whether it was easier or more difficult to push blood in the veins away from the heart than it was to push blood toward the heart.

3. Use your observations to sketch the veins in your hand. Indicate the location of the valves.

Conclusion

4. Write a concluding statement about the direction of blood flow. Refer to the valves in the veins of your hand.

Blood Pressure

During your last visit to a doctor, you may have noticed an apparatus for measuring **blood pressure** hanging on the wall. The gauge may resemble a thermometer; but instead of measuring temperature, this gauge is designed to measure pressure in terms of the height that a column of mercury can be raised. The greater the pressure, the higher the column of mercury rises in the tube. This is the origin of **millimetres of mercury**, the traditional unit for measuring blood pressure. The symbol for this unit is **mmHg**.

blood pressure: the pressure exerted by blood against the walls of blood vessels such as arteries

millimetres of mercury: a unit for measuring pressure in terms of the height of a column of mercury that can be supported by that pressure

mmHg: the symbol for millimetres of mercury

The term *blood pressure* usually refers to the pressure exerted by blood on the walls of a major artery. As shown in Figure A1.7, this is an indirect measurement in which the arterial blood pressure is equal to the pressure of the air in an inflatable cuff around the patient's arm, which is then equal to the pressure exerted by a column of mercury.

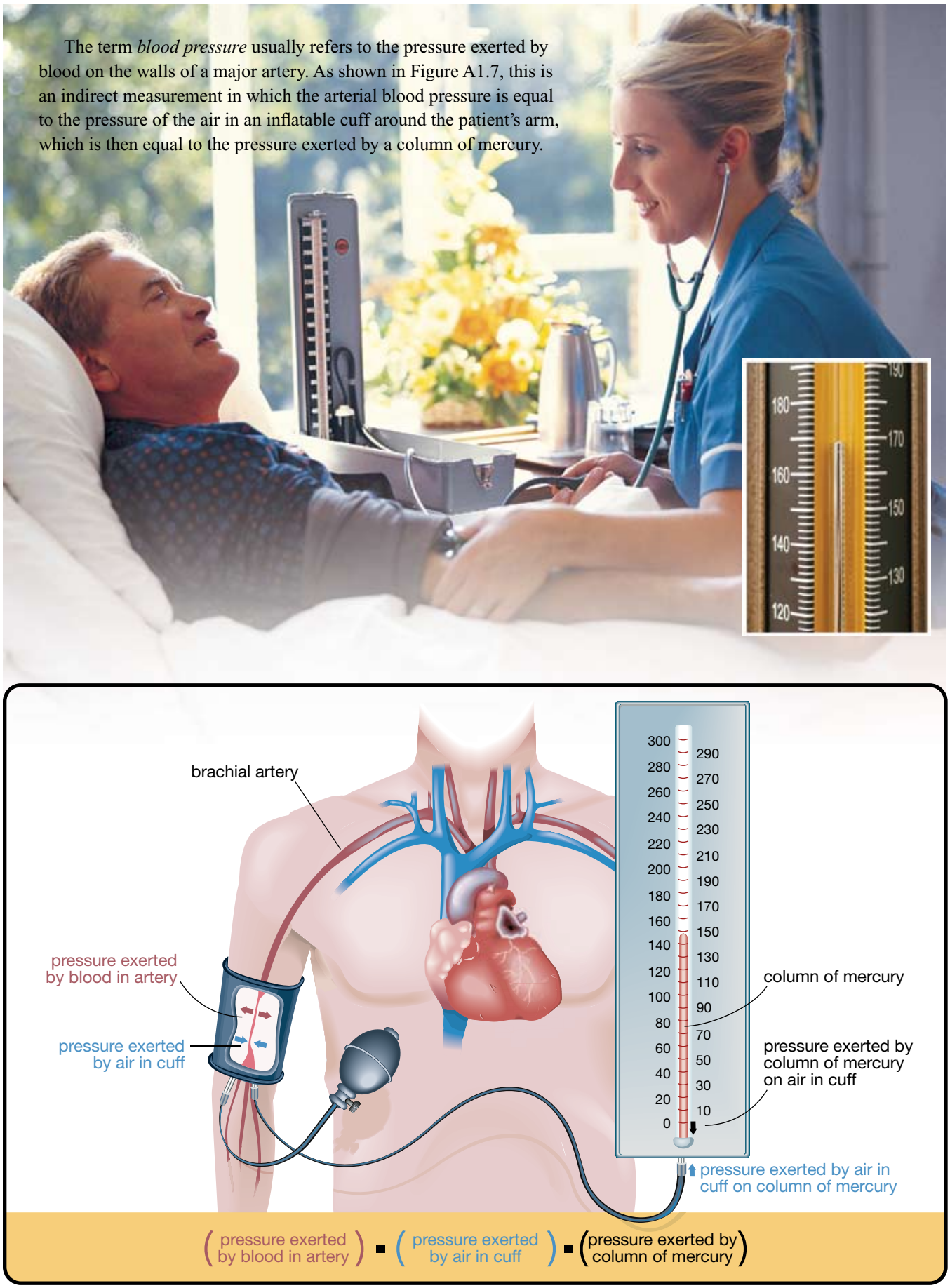


Figure A1.7: The pressure in the column of mercury is equal to the pressure of the blood in the artery.

Blood pressure forces the blood to flow through the body's vessels. Since the heartbeat has a cycle of contraction and relaxation, two pressures are measured with blood pressure.

The first number in a blood pressure reading is the larger **systolic pressure**, which represents pressure in the arteries when the heart's ventricles are contracting. The elastic fibres surrounding the arteries stretch slightly in response to this pressure.

systolic pressure: the pressure exerted on the artery walls when the heart's ventricles are contracting

diastolic pressure: the residual pressure exerted on the artery walls when the heart's ventricles are relaxing

The second number, called **diastolic pressure**, is smaller and represents the residual pressure in the arteries when the heart's ventricles are relaxing and the chambers of the blood are refilling. This pressure is due to the elastic walls of the arteries attempting to return to their previous shape between ventricle contractions.

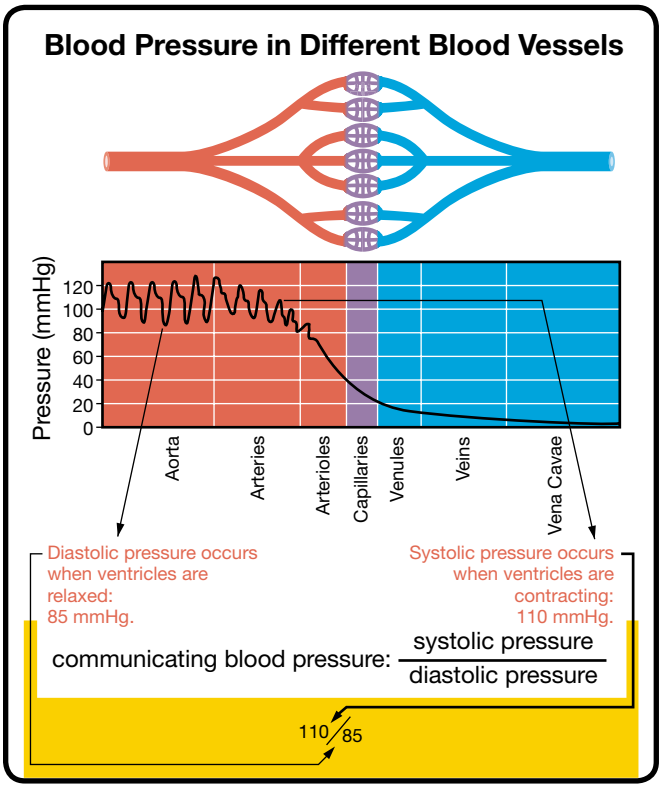


Figure A1.8

Blood pressure is written as systolic pressure over diastolic pressure. Using the values in Figure A1.8, the blood pressure would be 110/85. Even though this blood pressure value is read as *110 over 85*, this form of communication is not a fraction, so it should not be simplified or reduced. Note that the units for each individual pressure value are recorded in millimetres of mercury, but no units are recorded when communicating systolic pressure over diastolic pressure: each value is understood to be in millimetres of mercury.

The normal range of blood pressure for adults is a systolic pressure between 90 and 135 mmHg with a diastolic pressure between 50 and 90 mmHg. Blood pressure values in excess of 140/90 are considered to be high blood pressure or **hypertension**.

hypertension: chronic, abnormally high blood pressure, characterized by values greater than 140/90

Note that by the time blood leaves the arterioles and then enters the capillaries, pressure is significantly reduced. Since the smallest capillaries only allow blood cells to pass in single file, there is more resistance to the blood flow. Therefore, there is a reduction in blood pressure. So, how does blood return to the heart? Recall that the skeletal muscles squeeze the veins during exercise. This is combined with the action of one-way valves within the veins to force blood back to the heart.

Pressure and Blood Flow

As water flows from an outside faucet through a garden hose to a sprinkler or a nozzle, pressure on the water drives it through the hose. If there are no leaks, then the number of litres per minute that leave the hose should equal the number of litres per minute that enter the hose. Although this appears to be stating the obvious, it explains the behaviour of water as it leaves the hose. As an example, consider what happens when you clamp your thumb over the open end of a hose. Why does the flow become a jet-like spray?

If you put your thumb over the end of a hose, you reduce the cross-sectional area of the opening. The small opening forces the water to leave much faster to balance the number of litres per minute that enter the hose at the faucet end. Similarly, if the attachment on the hose's end increases the cross-sectional area, then the speed of the water drops since the larger opening easily accommodates the number of litres per minute entering the hose.

How does this thinking apply to the flow of blood from arteries to capillaries? Even though each individual capillary has a very tiny cross-sectional area, the huge number of capillaries fed by an artery means that the total cross-sectional area is much greater. One result of this is that the speed of the blood drops dramatically as it passes through a capillary bed, as in Figure A1.9. Another result is that the increase in cross-sectional area also contributes to the drop in blood pressure as the blood flows through a capillary bed from the arteries and arterioles.

The fact that blood travels slowly through the capillary bed means that the exchange of substances through diffusion between cells of tissue and the blood is enhanced. As blood leaves the capillary beds, the total cross-sectional area of the vessels decreases. As a result, the blood flow speeds up. However, because blood pressure is so low by the time it leaves the capillary beds, the flow speed through veins is much less than the speed through arteries.

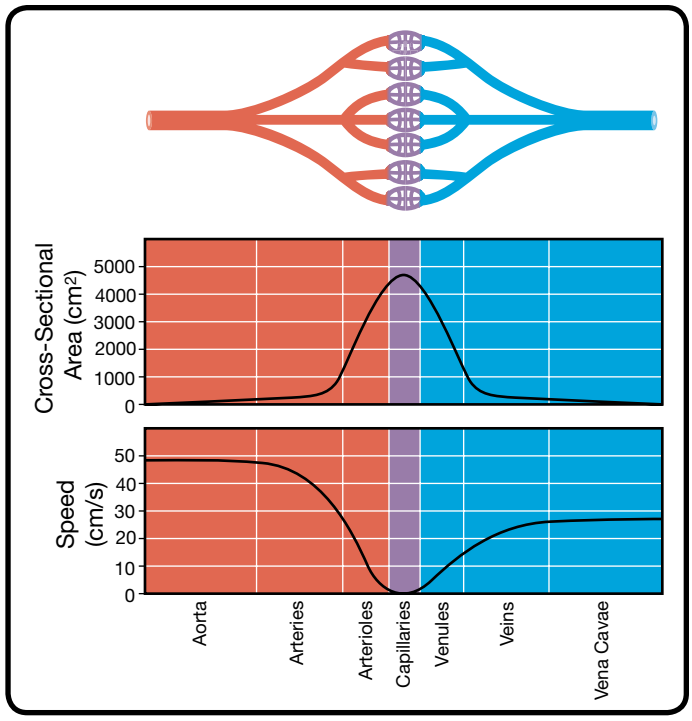


Figure A1.9

Practice

20. While waiting at a pharmacy to pick up a prescription, you decide to have your blood pressure tested using the automated machine available for customers. The machine says that your blood pressure is 138 over 96.

- **a.** Explain what the values of 138 and 96 measure. What is happening in your heart and arteries?
- **b.** Identify what unit could be included with each measurement you explained in question 20.a.
- **c.** Is 138 over 96 a cause for concern? What would you do with this information?

21. During diastole, the heart's ventricles are relaxing but there still is residual pressure in the arteries. Identify the source of this pressure.

22. Explain what causes the blood flow velocity to drop as it passes through the capillaries.

Measuring Blood Pressure

Blood pressure readings are often taken as part of a regular medical checkup. If blood pressure is too high, there is a risk of blood vessels bursting. This would be particularly dangerous if a vessel burst in your heart or brain. If blood pressure is too low, not enough blood can get to all the vital parts of your body. This may cause dizziness or fainting.

Your body has mechanisms to help control the amount of blood pressure. If your blood pressure is low, the blood vessels will be constricted or narrowed. If your blood pressure is high, the blood vessels will be dilated or widened. Many factors affect blood pressure. Readings can vary greatly between individuals due to the strength or rate of heart contractions or the elasticity of arteries. Higher blood pressure readings can also be attributed to anxiety level, exercise, a greater than normal amount of blood in the vessels, viscosity (thickness) of the blood, kidney disease, the presence of chemicals—including caffeine—in the body, or the narrowing of blood vessels due to a buildup of plaque along artery walls.

Investigation

Measuring Blood Pressure

Purpose

You will measure your blood pressure while resting and immediately after exercise.

Science Skills

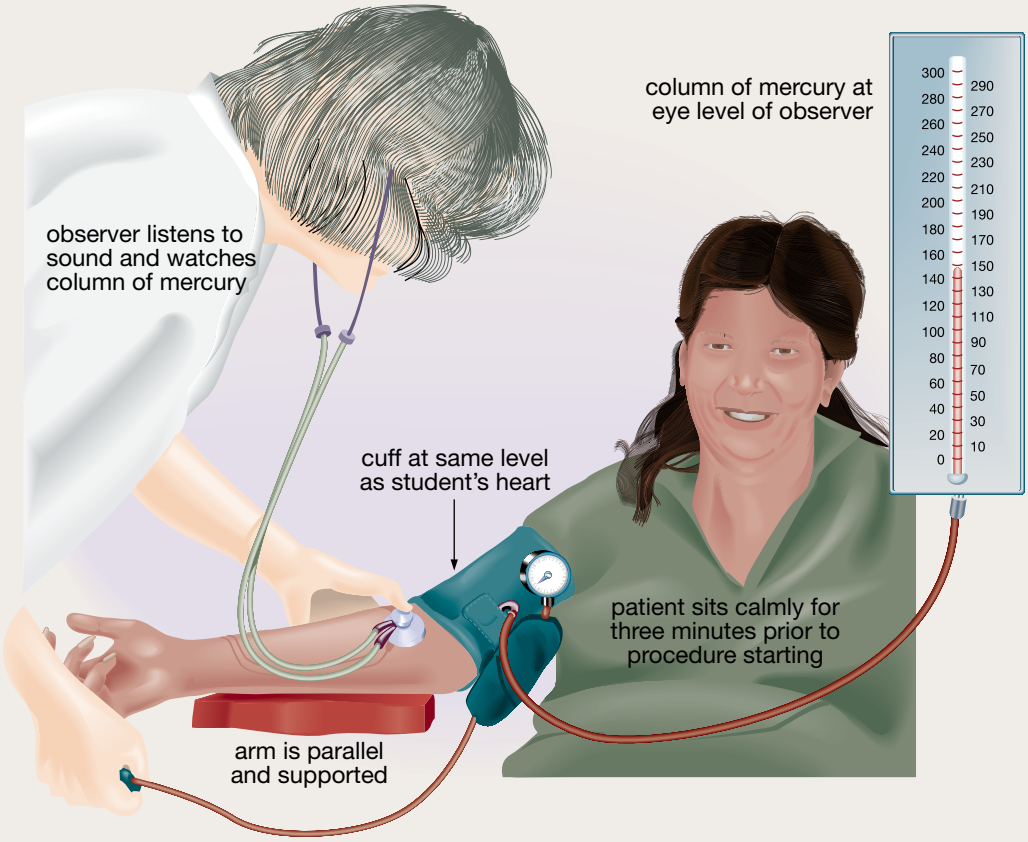
- ✓ Performing and Recording
- ✓ Analyzing and Interpreting

Background Information

To assess your health, a doctor or nurse may measure your blood pressure using either an automated digital machine or an older type of instrument called a **sphygmomanometer**. To use a sphygmomanometer, the person measuring your blood pressure inflates a cuff around your arm and listens to the sounds of your arteries with a stethoscope. The instant certain sounds change, the height of the column of mercury is noted for that moment.

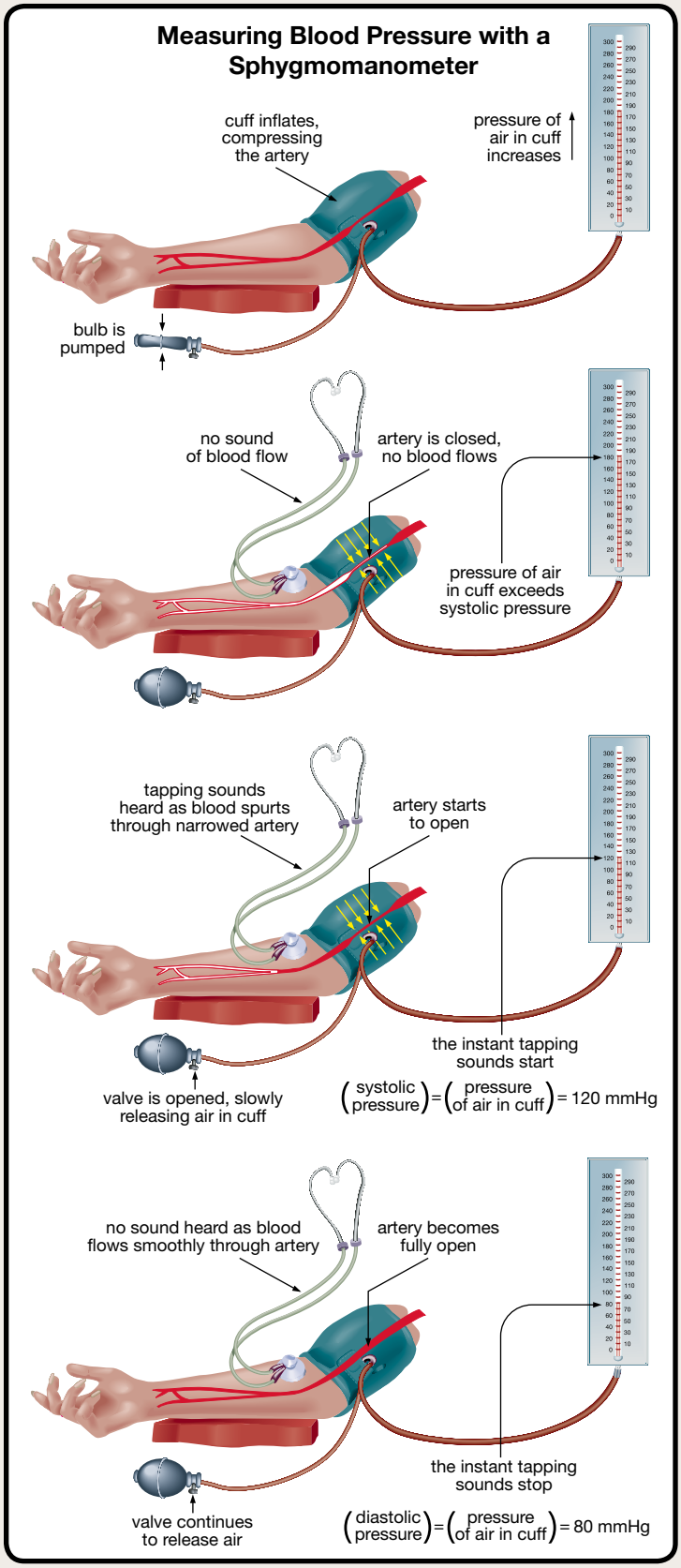
The steps for measuring blood pressure are outlined in the following illustrations.

sphygmomanometer: an instrument for measuring blood pressure



CAUTION!

- Prior to starting this investigation, be sure to carefully read the provided investigation instructions and those details included with the machine that you will be using.
- **Do not exceed 160 mmHg when inflating the cuff on the sphygmomanometer.**
- Remember that only a health-care professional, such as your doctor, can diagnose an abnormality in your blood pressure. A higher than average reading in this investigation is not necessarily an indication of high blood pressure.
- Mercury is a hazardous substance that can produce serious negative health effects. If a sphygmomanometer breaks and mercury spills into the open, the substance must be cleaned up immediately and thoroughly using proper procedures.
- If you have a medical condition that prevents you from participating in physical education classes, you should not participate in the exercising part of this investigation.



Procedure

step 1: You should be rested and sitting comfortably before beginning this activity. Using either a digital blood pressure machine or a sphygmomanometer and a stethoscope, have a classmate take your resting blood pressure. Record this number (in mmHg) as your resting blood pressure. If you use a digital machine for measuring blood pressure, follow the instructions provided with that machine. If you do not have access to a sphygmomanometer or a digital machine, measure your blood pressure by visiting a local pharmacy that has an automated blood pressure machine available for customer use.

step 2: Engage in four minutes of physical activity (jumping jacks, running on a spot, stepping up and down from a chair or stool) and have your blood pressure taken again at the end of the four minutes. Be sure that each class member performs the same exercise for the same amount of time for this activity.

Analysis

1. Obtain a class average for resting blood pressure. Compare the class average to the average adult blood pressure of 120 mmHg/80 mmHg. Describe how your own resting blood pressure compares to the average adult blood pressure.
2. Compare your blood pressure before and after exercising. Explain why your blood pressure changed after the exercise.
3. Compare the change in your systolic blood pressure reading after exercise to the change in your diastolic blood pressure reading after exercise. Did the readings change by the same amount? Can you account for the changes observed?
4. List some sources of error that may have affected the accuracy of the measurements made in this activity. Describe some improvements that could create more accurate measurements.

In your health file, record your resting blood pressure level and your blood pressure level after exercise.

Investigation

Blood Pressure and Heart Rate

Purpose

You will design an experiment to investigate a factor known to have an effect on blood pressure and heart rate.

Science Skills

✓ Initiating and Planning

Background Information

This investigation will allow you to apply what you have learned so far about blood pressure, heart rate, and the circulatory system. You have already been introduced to several factors known to have an effect on both blood pressures and heart rates.

Choose one of these factors. Then design an experiment that will allow you to test the effect of this factor on both blood pressure and heart rate.

You may decide to undertake some background research on the factor that will be the focus of your experiment. This will help you generate questions and identify what kind of data you will be collecting. It is also useful to research the importance of establishing a double-blind test when designing your experiment.

Process

The end product will be a detailed procedure for an investigation. You'll describe how to complete the necessary measurements and observations.

Procedure

step 1: Identify a specific question that needs to be investigated to determine the effects of the variable you have chosen to study.

step 2: Identify the manipulated variable, the responding variable, and the control group for your experiment. Based upon your background research, define a double-blind test and relate it to how the data will be collected in this experiment.

step 3: Determine what data needs to be collected to answer the question identified in step 1. Describe a means to collect that data, and list the tools required. Be sure to include any necessary safety precautions.

step 4: Design and construct data tables to ensure all the necessary observations are made and recorded.

1.2 Summary

The circulatory system's basic components are the heart, the blood vessels, and the blood. In this lesson you learned that vessels in the circulatory system are defined by their size and the direction in which they carry blood, relative to the heart. The vessels are specialized for their specific functions. Capillaries are uniquely designed for the exchange of nutrients between the body's cells and the circulatory system. Because matter exchange between capillaries occurs by diffusion, every cell in the body must be close to a capillary.

The pumping of the heart's ventricles exerts pressure on blood, and this pressure is then transferred to the artery walls. Blood pressure has two readings. The systolic reading is the artery pressure when the heart's ventricles are contracting. The diastolic reading is residual artery pressure when the heart's ventricles are relaxed. When listed separately, the units of millimetres of mercury are included with each of these pressure values. When communicated together, the units are usually omitted and the pressures are communicated as systolic pressure over diastolic pressure. An average blood pressure reading for adults is 120/80. Readings of 140/90 or greater are considered to be high blood pressure or hypertension.

Blood pressure is greatly reduced as the blood flow encounters resistance when passing single file through the many kilometres of tiny capillaries. By the time blood passes to the veins, blood pressure is so low that the blood is helped back to the heart by one-way valves and the contractions of skeletal muscles.

1.2 Questions

Knowledge

1. Copy and complete the following table to compare arteries, veins, and capillaries.

Characteristic	Arteries	Veins	Capillaries
description of vessel walls			
direction of vessel blood flow in relation to heart			
blood oxygen level in vessel			
colour in a circulatory system diagram			
blood pressure in vessel			
valves present			
pulse present			

Applying Concepts

2. People who have type 1 diabetes do not produce insulin—the sugar-regulating hormone—and they must have regular hypodermic insulin injections to regulate their blood sugar. Researchers are working on developing a dry powdered form of insulin that can be delivered by the same kind of inhaler used by people with asthma.

- **a.** Describe some possible benefits of the inhaler delivery system.
- **b.** Insulin is usually injected into fat underneath the skin. List the pathway that injected insulin takes from a capillary bed under the skin to a target cell in the liver.
- **c.** List the pathway that inhaled insulin would take from the lungs to a target cell in the liver.
- **d.** Which of the two delivery methods—injected or inhaled—would be faster at getting to target cells?

3. Identify some factors that can cause a person's blood pressure to increase.

4. Explain why it is more dangerous if an artery—rather than a vein—is cut in an accident.

5. Sketch a capillary bed. Include the artery, the arteriole, the vein, the venule, and the proper placement of valves. Include a few tissue cells being fed by the capillaries. Add arrows to your sketch that indicate the direction of blood flow, and add arrows that show what materials are being exchanged and the exchange direction.

6. Soldiers on guard are often required to stand in one place for long periods of time. While standing at attention some of the soldiers sway back and forth, slightly contracting and relaxing their calf muscles. Other soldiers exercise the muscles in their lower legs by slightly wiggling their toes in oversized boots. Soldiers who do not use strategies like these often faint after standing for a long time. Explain why contracting and relaxing the muscles in their lower legs helps prevent soldiers from fainting.

1.3 Blood

Your school is organizing a blood drive for the local blood bank. If you have not given blood before, you probably have questions about what will happen during the donation. Before blood is donated, potential donors—aged 17 or more—are asked a list of questions about their general health, their travel history, and their participation in certain activities. The answers determine if there is a risk that the donor's blood carries a disease that could be passed on to the person receiving the blood. In addition to medical concerns, some people are prohibited from donating blood or receiving transfusions for religious reasons.

If you are able to participate, a nurse will test your temperature and blood pressure to see if you are healthy enough to donate. The nurse will also take a drop of blood from your finger and time how long it takes for the blood to sink in a solution to examine the oxygen-carrying capacity of your red blood cells.

The nurse will then insert a sterile needle into a vein in your arm and take about 450 mL of blood. Your body contains about 5 L of blood, and this small donated volume of 450 mL will quickly be replaced by new blood cells formed in the spongy marrow inside your bones. Since the donated blood can become thick and clot quickly in the air, a chemical called an anticoagulant must be added to stop the collected sample from clumping.

The collected blood will then be taken to a lab where it will be tested to ensure it is free of dangerous diseases, such as hepatitis and HIV. Once this screening process is complete, the donated blood may be given to patients who have a need for the entire sample or some of its components.

It may be donated as whole blood, but later the individual parts of blood may be separated. Some patients require the portion that causes clotting, while others are given only the liquid part of blood to replace fluids they have lost in an accident or during major surgery. It is estimated that one blood donation is able to help three other people, and it quite often helps save lives.

Blood is the fluid of life. Because the jobs performed by blood are so important, people die if they lose too much of this vital fluid. Since the average adult only has about 5 L of blood, it must be quickly cycled by the circulatory system to efficiently carry out its jobs. In previous lessons you learned that blood is constantly circulated throughout the body so it can deliver oxygen and nutrients and remove wastes from cells as they carry out their functions—however, blood also transports other materials such as chemical messengers (called hormones), vitamins, and minerals. Blood also helps to protect people's bodies from disease and maintains water balance, temperature, and pH. Health-care workers examine blood samples from people who are not feeling well because irregularities in a blood sample can help diagnose an imbalance in the body.

Try This Activity

A Circle Diagram for Blood

So many images come to mind when we think about blood because we are aware of the vital importance of this fluid flowing within our bodies. Just the sight of lost blood makes some people uncomfortable. Blood is often used as a symbol of our spirit and can signify both life and death. Children are someone's "flesh and blood," murderers have "blood on their hands," something that you were meant to do is "in your blood," and hard work requires "blood, sweat, and tears."

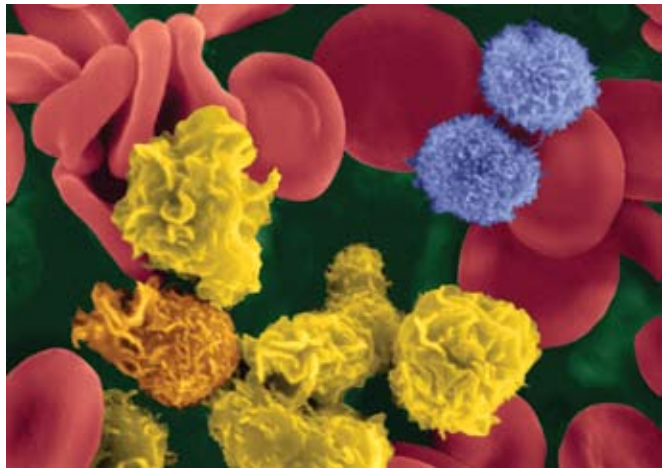
Place the word *blood* within a small circle at the centre of a blank page. Draw a larger circle around the smaller one. Brainstorm all the different ideas, terms, metaphors, symbols, and sayings that come to mind with this word and write them within the larger circle. Draw a box around the large circle. Determine the sources for ideas and terms that you wrote within the larger circle, and write these terms within the box. As an alternative, this activity may be done in small groups or as a class, with many students contributing to the formation of one circle diagram.

Blood Components

Blood appears to be a liquid with a uniform red colour, but blood is actually a mixture of living cells and pieces of cells suspended in a broth-like liquid. If you let a blood sample sit for a while or spin a blood sample in a machine called a centrifuge, the blood separates into layers to reveal its different parts.

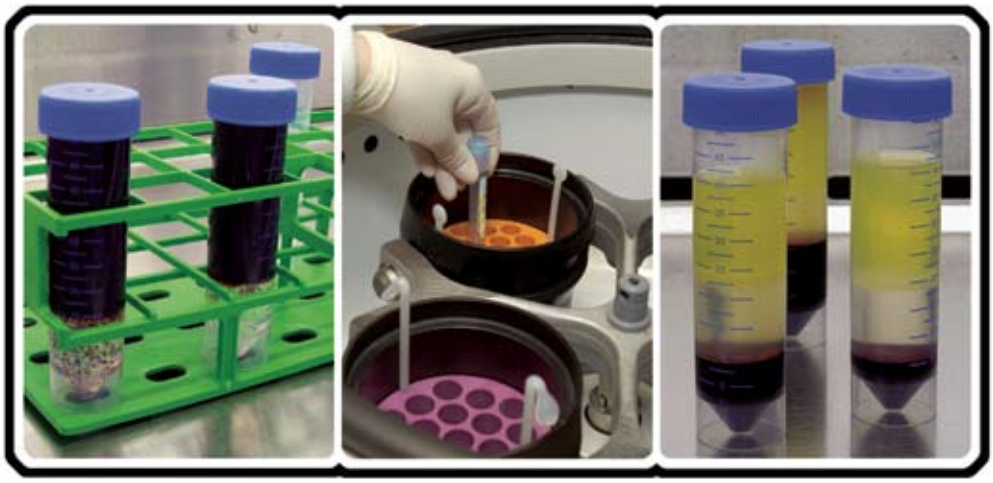
At the bottom of the centrifuge sample, the red blood cells are clearly visible. The pale yellow liquid that is seen to occupy the top half of the blood samples is called **plasma**.

Plasma is like the broth of a soup because it is mostly water with substances dissolved or floating in it. About 55% of blood volume is due to the watery plasma. In between the plasma and the red blood cells is a section of clear fluid containing the **white blood cells** and **platelets**. Platelets contribute the smallest amount to the volume of blood.



White blood cells are designed to protect the body from disease-causing organisms and other harmful materials. Sometimes these foreign substances enter the body through a cut in the skin. At the site of a cut, the platelets are tiny fragments of cells that play a key role in helping blood clot. The clot eventually hardens to form a scab that keeps the wound clean while new replacement skin grows. In this lesson you'll learn more about the roles played by plasma, red blood cells, white blood cells, and platelets.

Separation of Blood into Components



- **plasma:** the pale yellow fluid portion of blood where the cells are suspended
- **white blood cell:** a colourless blood cell that acts to defend the body against diseases and other foreign invaders
- **platelet:** a particle found in the bloodstream that begins the blood-clotting process at the site of a wound

Practice

Obtain a copy of the handout "Separating Blood into Its Components" from the Science 30 Textbook CD. Use this handout to answer questions 23 and 24.

23. Add the following labels to this diagram: red blood cells; plasma; white blood cells; platelets; sample of blood prior to placement in centrifuge; sample of blood after removal from centrifuge; and centrifuge.

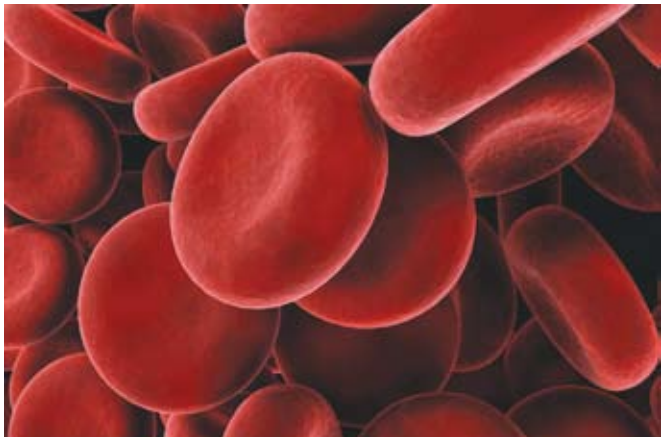
24. Identify the blood component(s) that best describes each of the following statements.

a. This component comprises about 55% of blood volume.

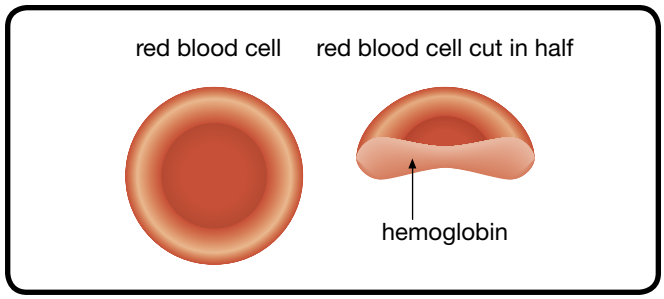
b. This component makes up almost 45% of the total volume of blood.

c. Together, these components comprise less than 1% of the total volume of blood.

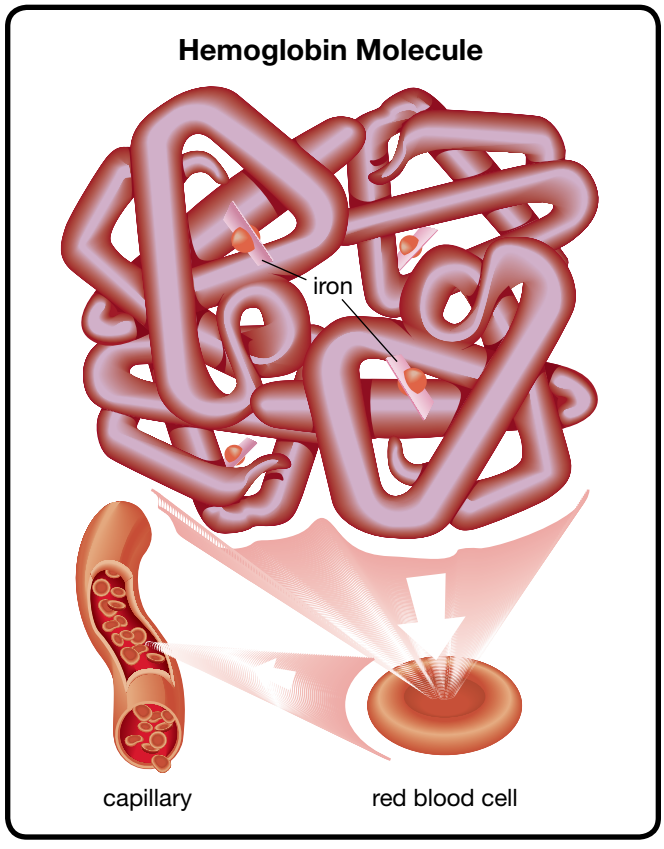
Red Blood Cells



Red blood cells, also called **erythrocytes**, are the most numerous cell type in a blood sample. In one drop of blood there are about one million red blood cells. These cells are designed to transport oxygen. They are shaped like a covered inner tube, with a depression in the middle but not a hole. This distinctive shape—called **biconcave**—allows them to slide through the blood vessels with ease. If their shape were more square or jagged they might stick to each other or the vessel walls, which would result in a jam that slows or stops circulation in that vessel. The cream-filled, doughnut-like shape provides a large surface area to volume ratio for an efficient gas exchange.



A red blood cell's lifespan in the bloodstream is about 120 days. As red blood cells die, they are absorbed by the liver and are soon replaced with new cells produced in the bone marrow. The bone marrow produces millions of red blood cells every second. Each red blood cell takes about two days to develop. As the red blood cells mature and are released from the bone marrow, they lose their nucleus. Having no nucleus provides extra room to pack the cell full of the **hemoglobin** molecule.



Hemoglobin is a pigment that gives the red blood cells and whole blood their red colour. It contains iron which interacts with the oxygen present in the lungs. A hemoglobin called **oxyhemoglobin** is bound to oxygen and has a bright red colour.

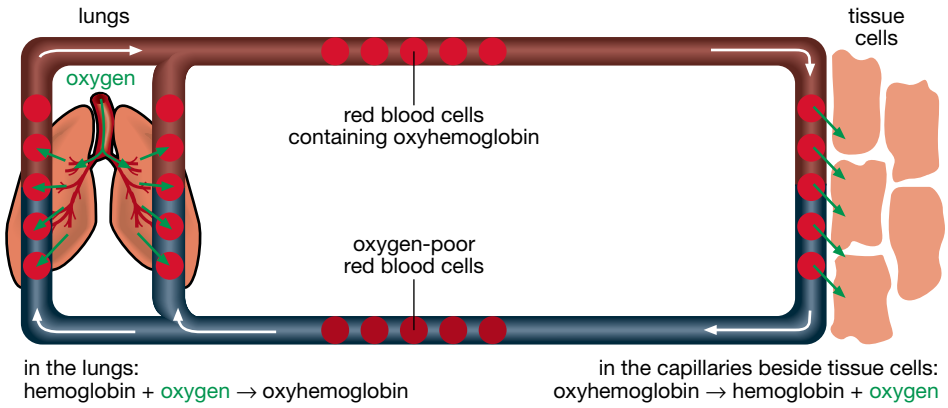
erythrocyte: a term for a red blood cell that contains hemoglobin and transports oxygen from the lungs to the body's cells

biconcave: the distinctive shape of red blood cells where the cells are flat but dip inwards at the centre on both the top and bottom

hemoglobin: an iron-containing pigment that binds oxygen to facilitate its movement in the circulatory system

oxyhemoglobin: a hemoglobin bound with oxygen that appears bright red in colour

As red blood cells pass through the lungs, hemoglobin molecules pick up oxygen molecules to form oxyhemoglobin. The red blood cells are transported through arteries, arterioles, and eventually to capillary beds next to body tissues. As these cells slowly pass through capillaries near tissue cells, the reaction is reversed: the oxygen is released and the blood loses its bright red colour.



Investigation

Iron-Fortified Cereals

Background Information

New red blood cells are constantly being made to replace old red blood cells, and iron is needed for the production of the red blood cell's hemoglobin. Good sources of iron include foods such as meat, fish, poultry, beans, dried fruits, and whole grain breads. For people who don't receive enough iron through their regular diet, it is beneficial for them to supplement their iron supply by eating foods with added iron. Many cereal companies advertise that their product is "fortified with iron."

Purpose

You will separate the iron from breakfast cereal enriched with iron.

Materials

- 750 mL (3 cups) of iron-fortified cold breakfast cereal
- mortar and pestle or a rolling pin
- two 1000-mL beakers
- magnetic stirrer complete with magnetic stir bar and magnetic retrieval wand
- 700 mL of distilled water
- plastic rinse bottle with distilled water

Procedure

step 1: Crush the cereal with a rolling pin or a mortar and pestle until the cereal becomes a fine powder. Place the powdered cereal in one of the 1000-mL beakers.

step 2: Add 700 mL of distilled water to the other 1000-mL beaker.

step 3: Carefully add the stir bar to the beaker's bottom and place the beaker on the magnetic stirrer. If the magnetic stirrer is combined with a hot plate, do not turn on the hot plate.

Science Skills

✓ Performing and Recording
✓ Analyzing and Interpreting

step 4: Turn on the magnetic stirrer at a moderate speed setting. While it is spinning, carefully add the powdered cereal. The rotation speed may have to be increased.

step 5: Stir for approximately 30 minutes.

step 6: Turn off the magnetic stirrer. Use the retrieval wand to carefully remove the stir bar. Gently rinse any cereal off the stir bar and then observe its surface.

Analysis

1. State the iron property that accounts for what you observed on the magnetic stir bar's surface.
2. People who don't get enough iron from their diets, or those people who suffer from chronic blood loss, have lowered red blood cell levels and can develop a blood condition called *anemia*. Based on your knowledge of red blood cell functions, what symptoms do you think people with anemia might have?
3. One test used to see if potential blood donors have anemia is to put a drop of a donor's blood in a vial of thick blue fluid. If the blood drop falls very slowly or does not sink at all, it could be a sign that the donor is anemic or low in red blood cells on that particular day. Explain why a sample of blood from someone who has anemia might fall less quickly than a sample of blood from a person who does not have anemia.
4. To stay healthy, men require about 10 mg of iron per day and women about 15 mg per day. Pregnant women daily need about 30 mg of iron. Explain why pregnant women might require double the mass of iron required by women who are not pregnant.

Science Links

The interaction of iron objects with a magnet played a key role in "Iron-Fortified Cereals." In Unit C you will continue to explore these effects by studying the interaction between magnetic effects and electrical effects.

White Blood Cells

White blood cells, also called **leukocytes**, are much larger than red blood cells and are found in much lower numbers. In a healthy individual there is only about one white blood cell for every 600 to 700 red blood cells. Like red blood cells, leukocytes are made in a person's bone marrow. Unlike red blood cells, leukocytes keep their nucleus when they mature. Their shape is not uniform and they lack a colour pigment. In prepared slides and photographs, white blood cells are often stained so that they can be observed more clearly. The life span of a white blood cell is generally about 13 to 20 days.

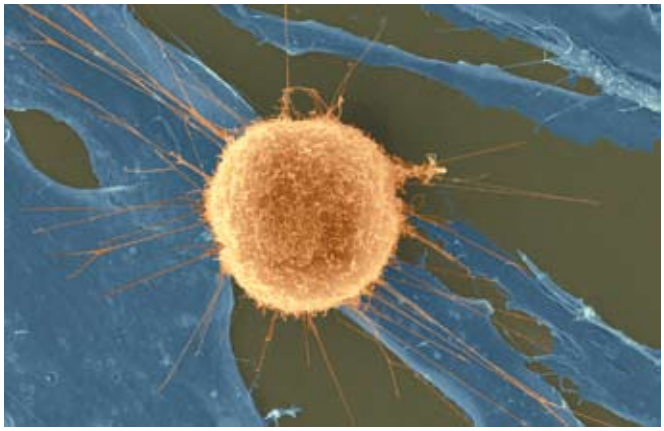


Figure A1.10: A human white blood cell attacks cancer cells.

When disease-causing organisms enter the body, the number of white blood cells increases by releasing stored white blood cells from the body or producing more in the bone marrow. A doctor will often request that a white blood cell count be performed in a blood sample. Elevated white blood cell counts usually signal an infection.

- **leukocytes:** a term for white blood cells
- **protein:** a large organic molecule consisting of a chain of amino acids; an essential building block of all cells that plays a key role in the functioning of body systems
- **fibrinogen:** a soluble protein present in blood plasma that converts to fibrin when blood clots

Practice

25. Cancer patients often experience extreme fatigue because the cells in their muscle tissues are oxygen depleted. This condition exists because the chemotherapy many patients undergo to treat their cancer has a side effect of reducing the number of blood cells.

- **a.** In these circumstances, identify what type of blood cells are especially needed by cancer patients.
- **b.** The type of blood cells in question 25.a. must be separated from whole blood. Describe a process that could separate whole blood into its components.

26. The body produces millions of red blood cells in the bone marrow every second. White blood cells are made in bone marrow at twice the rate of red blood cells. However, in a given sample of blood, nearly 45% of the blood consists of red blood cells and less than 1% is made of white blood cells.

- **a.** Suggest a reason for the lower volume of white blood cells in a sample of blood even though they are produced at twice the rate of red blood cells.
- **b.** Most agents that cause disease are outside the bloodstream and are found in the fluid spaces between tissue cells. Use this information to develop another reason for the lower volume of white blood cells in a sample of blood.

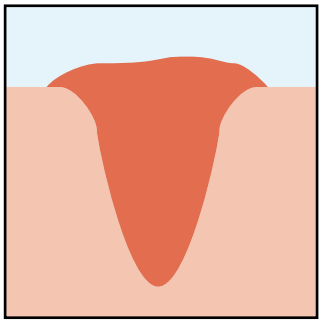
Platelets

Think back to the last time you had a small scrape or a minor cut to your skin and the tissues below your skin. The bleeding usually stops after a few minutes because **proteins** in your blood work to form a plug that seals the damaged blood vessels. A protein present in blood and capable of forming a plug is called **fibrinogen**. This protein remains inactive until called into action by the platelets.

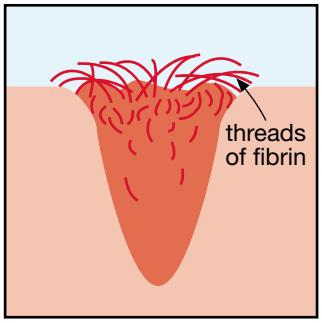
Platelets rupture when they come into contact with a rough surface, and they trigger a complex series of chemical reactions. These reactions cause the dissolved fibrinogen protein to convert to its active form—a thread-like protein called **fibrin**. The fibrin threads become interwoven to produce a mesh that traps red blood cells. As more red blood cells get caught in the net of fibrin, the combination of these fibres and blood cells produce a thick red jellylike substance called a **blood clot**. When this mesh of threads and red blood cells hardens and dries, it is called a scab. The skin under the scab heals and the scab eventually falls off.

The Clotting Process

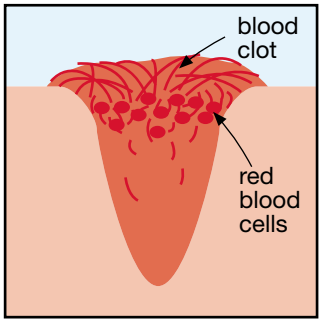
The skin is cut. Blood starts leaking out of the body to wash out dirt and germs from the cut.



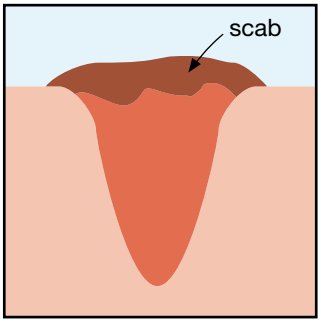
Platelets come into contact with the rough surface of the cut. The platelets rupture and release chemicals that convert fibrinogen into threads of fibrin.



Red blood cells get caught in the fibrin net to form a blood clot.



The clot hardens to form a protective barrier known as a scab.



Many life-threatening medical conditions are related to blood clotting. A diet high in fat can make the walls of arteries become rough—this causes platelets to adhere and produce blood clots inside the vessel. A blood clot in these circumstances can lead to serious cardiovascular problems, including heart attacks or strokes.

Some people lack the ability to produce the necessary blood proteins that allow platelets to form a clot. For these individuals, even minor cuts and bruises bleed excessively and take much longer to heal. This condition is called **hemophilia**. To keep healthy, people with hemophilia often need transfusions of protein clotting factors and platelets.

- **fibrin:** a thread-like insoluble protein formed from fibrinogen
  The threads of fibrin mesh to form the fabric of a blood clot.
- **blood clot:** a jellylike, solid mass consisting mainly of red blood cells trapped in a net of fibrin fibres
- **hemophilia:** a blood disorder involving the blood's reduced ability to clot, which can lead to excessive bleeding

DID YOU KNOW?

Alexei Romanov, the son of Nicholas II—the last Tsar of Russia—had hemophilia. At the time, there was little understanding and no treatment for the disease. Alexei's mother, Alexandra, employed the help of a self-proclaimed healer named Rasputin to help Alexei. Some people believed that Rasputin was able to use hypnosis to help control Alexei's condition and slow his bleeding. Rasputin was hated by many Russians for his powerful influence with the royal family, and he was killed by his enemies in 1916.

Investigation

Observing a Prepared Blood Smear

Science Skills

✓ Performing and Recording
✓ Analyzing and Interpreting

Purpose

You will use prepared slides of human blood to observe the structure and abundance of the cellular components of blood.

Materials

- prepared human blood slides
- compound microscope
- blank unlined paper
- pencil

Procedure

step 1: Review basic microscope skills by reading the handout "Using a Microscope" from the Science 30 Textbook CD.

step 2: Obtain a prepared human blood slide and view the slide under high-power magnification.

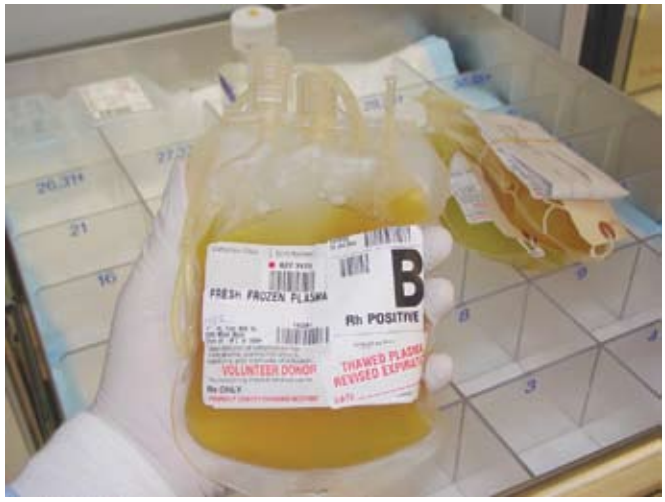
step 3: Identify and draw to scale a red blood cell, two white blood cells, and platelets—perhaps these are only barely visible. Remember that the blood sample is most likely stained for better visibility and, therefore, the colours may not be representative. Label the three cellular components of blood on your drawing. Include a label for the white blood cell nucleus.

step 4: Count and record the number of red blood cells and white blood cells in the field of view.

Analysis

1. Describe two physical differences between the observed red blood cells and white blood cells.
2. The function of red blood cells is to transport oxygen. How is a red blood cell's shape and structure related to its function?
3. Compare the two white blood cells that you sketched in step 3. Describe how these two cells are similar. Describe how they are different.
4. State the ratio of red blood cells to white blood cells in the section of the prepared blood slide that you observed.

Plasma



Plasma is a yellowish liquid that is like soup broth, because it is mostly water with substances dissolved or suspended in it. More than half of blood is composed of watery plasma. The plasma holds and transports such substances as the following:

- cells of the blood—red blood cells, white blood cells, and platelets
- dissolved waste carbon dioxide from the capillaries to be excreted out at the lungs
- urea, a waste product from the liver filtered by the kidneys to become urine
- hormones from glands
- digested nutrients such as glucose, amino acids, vitamins, and minerals from the digestive tract
- proteins such as fibrinogen that aid in blood clotting, as well as other proteins like antibodies involved in the immune response

When patients have lost a lot of blood due to an accident or major surgery, they may be given donated plasma to replace the lost fluid.

Practice

Use the following information to answer questions 27 to 30.

The majority of blood collected at a blood donation clinic is not left as whole blood. It is instead separated by a centrifuge into its components of red blood cells, plasma, and platelets. In this way, your single donation of one blood unit can help three other people.

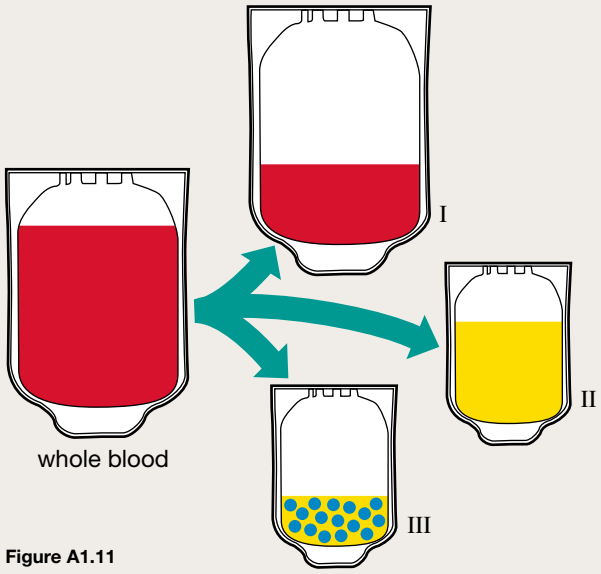


Figure A1.11

27. Refer to Figure A1.11. Identify the blood component that corresponds best to each of bags I, II, and III.

28. Burn victims suffer a significant loss of skin tissue and are, therefore, highly susceptible to deadly dehydration. What blood component best addresses this problem?

29. A cancer patient receiving radiation treatment is often unable to produce enough of a key blood component that prevents uncontrollable internal bleeding. Identify the blood component given to this patient in a transfusion.

30. Explain the following statement:

"Every time you give blood you can save up to three lives."

1.3 Summary

The typical human body contains about 5 L of blood that transports materials around the body to help prevent disease. Blood is a mixture of living and non-living components: red blood cells, white blood cells, platelets, and plasma.

The disc-shaped red blood cells are full of a molecule called hemoglobin, which allows these blood cells to carry and transport oxygen to the body's cells. White blood cells are designed to help protect the body from disease-causing organisms. Platelets are the blood parts that make blood self-sealing. When there is a cut, platelets form a clot to stop the loss of blood—people with hemophilia lack key factors in their blood that help form clots. Plasma is a yellowish liquid that the blood cells and platelets float in. Many substances are dissolved in plasma, including glucose, urea, and hormones.

1.3 Questions

Knowledge

1. a. List the four components of blood in decreasing order of their relative volume in whole blood (from most abundant to least abundant).

b. Sketch a scale diagram of each component of question 1.a.

2. Obtain a copy of the handout "Blood Smears" from the Science 30 Textbook CD. The first five blood smears on this handout come from a healthy patient. The sixth blood smear is from a patient with neutrophilia.

a. Identify the major difference between the blood smears from the healthy patient shown in slides 3, 4, and 5 with the blood smear from the patient with neutrophilia on slide 6.

b. Suggest a possible reason for the difference you identified in 2.a.

Applying Concepts

3. Carbon monoxide is a colourless, odourless gas produced during the combustion of fossil fuels, including gasoline. Carbon monoxide binds to the hemoglobin in red blood cells much faster and more strongly than does oxygen. Based on your knowledge of the role of red blood cells, explain why exposure to carbon monoxide can be so dangerous.

4. Leeches and vampire bats are both parasites that feed on animal blood. After they use their sharp teeth to cut the surface of the animal's skin, they release a blood-thinning chemical called an anticoagulant that not only stops blood from clotting but also allows greater blood flow by dilating blood vessels.

Explain why an anticoagulant might be useful for treating circulatory system problems.

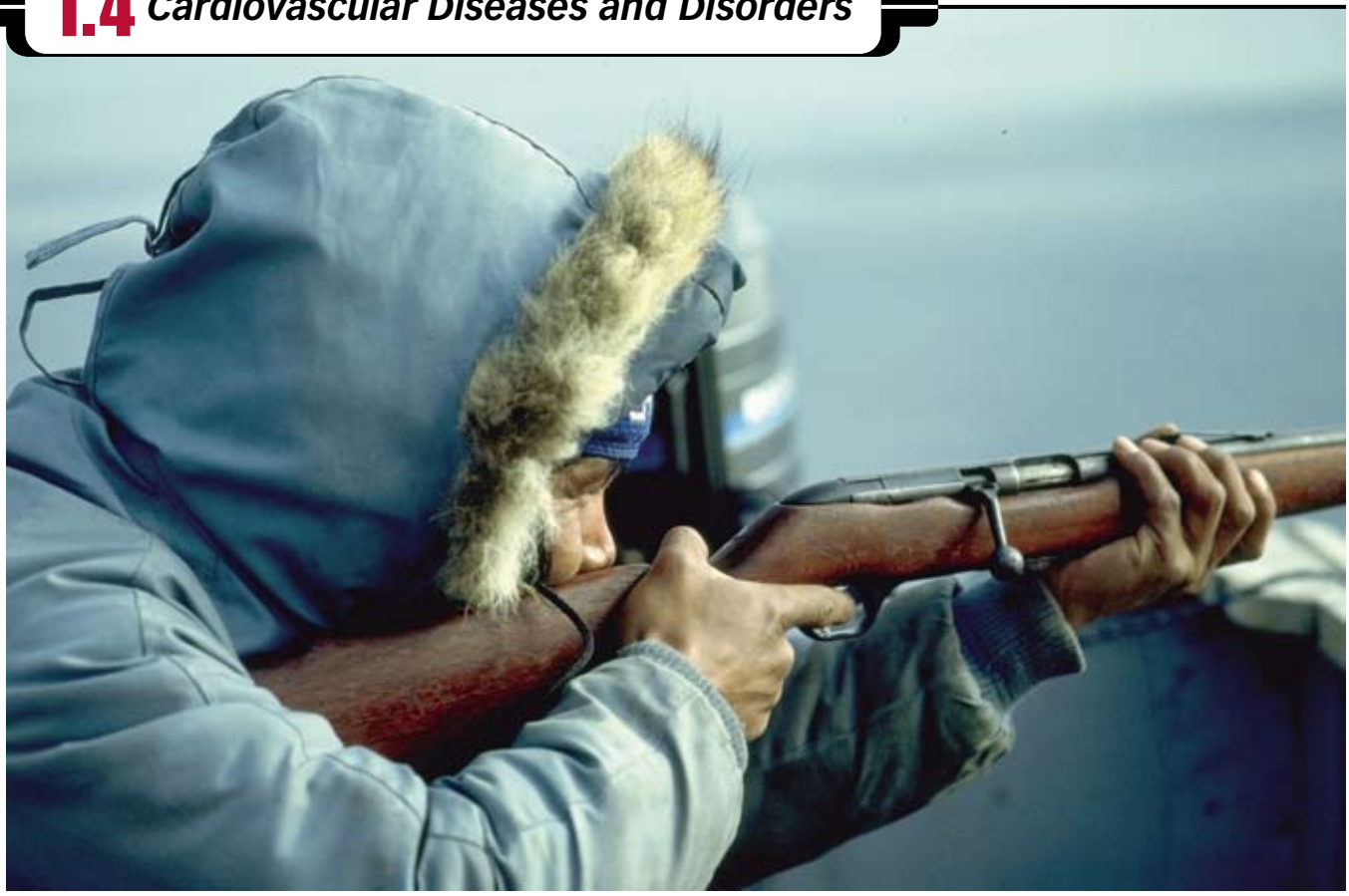
5. Leukemia is a type of cancer where the body produces large numbers of abnormal blood cells—particularly white blood cells—that do not function properly.

a. How could having improperly functioning white blood cells affect people with leukemia?

b. Why would doctors treat leukemia by giving patients a bone marrow transplant?

6. Do you meet the basic criteria to be a blood donor? Use the Internet to gather information about the basic eligibility criteria for donating blood in Canada.

1.4 Cardiovascular Diseases and Disorders



The traditional diet of Inuit people living in coastal communities has centred on fish like arctic char and marine mammals including seals, walruses, and whales. These trends even extend to traditional snack food. Maktaaq, which is whale skin attached to a few centimetres of insulating fat, or blubber, is considered to be one of the finest delicacies in the traditional Inuit diet because it is so delicious. Sometimes maktaaq is consumed immediately after the kill as the whale is being brought ashore, while other times the maktaaq and other whale foods are allowed to freeze and are then buried in snow-covered food caches for later use.

Nutritionists and medical researchers began to collect data in the 1970s about the eating habits and the incidences of illness and disease in traditional Inuit communities. The results of these studies were surprising. Even though fat from marine mammals was a staple of a traditional Inuit diet, the incidence of **cardiovascular disease** in the population was very low. This data stands in sharp contrast to data gathered from communities outside of the Arctic where a high-fat diet coincided with higher rates of cardiovascular diseases.

cardiovascular disease: one of many disorders—coronary heart disease, strokes, and varicose veins—that affect the heart and/or the blood vessels

Figure A1.12: Maktaaq is an Inuit delicacy.

If the Inuit diet is so high in fat, why is there such a low incidence of cardiovascular disease among Inuit who follow a traditional lifestyle?

The full answer is still under investigation, but as you will see in this lesson the preliminary findings are quite intriguing. To understand better the results of the early investigations, you will need to learn about the causes of some major cardiovascular diseases, the different types of fat that are part of your diet, and the **traditional ecological knowledge** of Canada's Inuit.

traditional ecological knowledge: the accumulated observations and understanding of the people living within an area, acquired over many hundreds of years through direct contact with the environment

Incorporating traditional ecological knowledge involves developing an understanding of human interactions with the environment and focusing on the inseparable relationship among land, resources, and culture.

Practice

In the 1700s, many Europeans who came to the Arctic to explore or hunt whales began to get a disease called scurvy after spending many months in the far north. Scurvy is a connective-tissue disease that is thought to be the number one cause of deaths at sea in the age of sail. Scurvy symptoms include pale skin, sunken eyes, tender gums, tooth loss, internal bleeding, and a physical and mental deterioration of the body that frequently leads to death.

It was eventually discovered that having fresh fruit and vegetables aboard ships prevented scurvy. The nickname of *limey* for a British sailor comes from the use of limes on British ships to prevent scurvy. It was not until 1932 that medical science established the biochemical cause of scurvy.

Meanwhile, Inuit people who lived their entire lives in the Arctic environment did not have access to fresh fruit and vegetables, but they did not suffer from scurvy.

Use the Internet to gather information to help you answer the following questions.

31. Identify the essential nutrient linked to the cause of scurvy. Explain why it affected sailors, explorers, and whalers on long voyages during the 1700s.

32. How did maktaaq play a key role in preventing scurvy within the Inuit population?

33. Prior to the use of limes and other fresh fruit, explain how the traditional ecological knowledge of the Inuit could have helped the early European explorers of Canada's Arctic who suffered from scurvy.

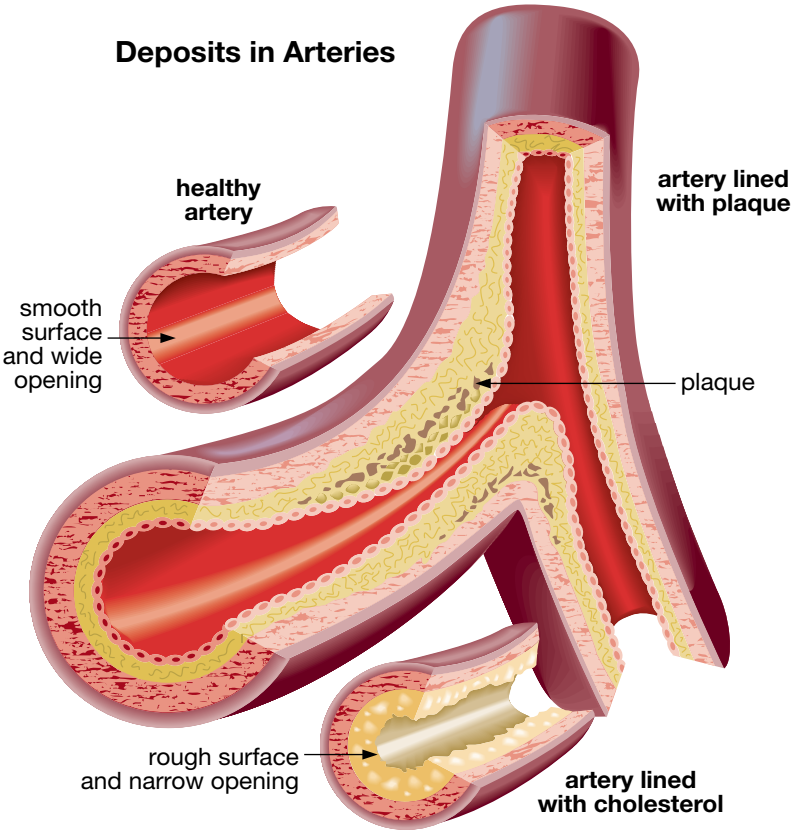
Cardiovascular Diseases and Cholesterol

As the terms *cardio* + *vascular* suggest, cardiovascular diseases are disorders of the heart + blood vessels. As shown in Figure A1.13, cardiovascular diseases include a wide range of disorders.

Cardiovascular Disease	Description of Disorder	Heart Disease	Blood Vessel Disease
atherosclerosis	hardening of arteries due to accumulation of fatty deposits		✓
coronary heart disease	restricted blood flow through coronary arteries resulting in chest pain and heart attack	✓	
heart attack	clot in a coronary artery cuts off blood supply to heart muscle and tissue dies	✓	
stroke	sudden loss of brain function caused by an interruption in blood flow to brain		✓
aneurysm	bulging or weakness in wall of artery or vein		✓
valvular heart disease	diseases of heart valves leading to narrowing, leaking, or improper closing of valves	✓	
septal heart defects	opening within septum that allows blood to flow between left and right ventricles of heart	✓	

Figure A1.13

Read through the descriptions of these disorders in Figure A1.13. Note that an interruption of the blood flow through key arteries plays a major role in many of the diseases on this table. In many cases, arteries lose their effectiveness because the blood has too high a concentration of **cholesterol**. Cholesterol is a vital component of all cell membranes and is a key ingredient in the production of important hormones and vitamin D. This essential substance creates problems when its concentration in the bloodsteam becomes too high. Accumulated cholesterol deposits build up along with fat and other debris on the inside walls of arteries. Like the sticky film of bacteria that builds up on your teeth before brushing, these sticky deposits in arteries are also called **plaque**. As plaque accumulates, the affected artery starts to narrow. Since the outer layer of plaque can harden into a rough and rigid surface, the artery becomes stiff. The narrowed opening and rough interior makes it more difficult for blood to flow.



cholesterol: a waxy, fat-like substance present in the cell membrane of every body cell and in food from animal sources

High levels of cholesterol can lead to cardiovascular disease.

plaque: a semi-hardened accumulation of substances originally suspended in a fluid

Cholesterol and fat are transported by special blood proteins in plasma through the bloodstream. About two-thirds of all the cholesterol in your blood can be found in **low-density lipoprotein (LDL)**. LDL is responsible for carrying cholesterol in the bloodstream from the liver to cells of body tissues. This is the cholesterol that may find its way to the inside walls of blood vessels and lead to plaque. You can see why these particles are often referred to as "bad cholesterol."

Another blood protein, called **high-density lipoprotein (HDL)**, carries cholesterol to your liver so it can be excreted from your body. Since this protein scours the bloodstream to collect excess cholesterol, it is sometimes called "good cholesterol."

- **low-density lipoprotein (LDL):** a blood protein that carries cholesterol in the bloodstream from the liver to the rest of the body

  Too much LDL in the blood leads to deposits on the walls of arteries, so this is referred to as "bad cholesterol."
- **high-density lipoprotein (HDL):** a blood protein that carries cholesterol in the bloodstream from the body cells to the liver

  High levels of HDL in the blood means it is less likely that deposits will form on the walls of arteries, so this is referred to as "good cholesterol."

Practice

34. If you were told to make a sandwich that contained no cholesterol, would you choose butter, cheese, ham, peanut butter, or tuna as a filling?

35. The following results come back from two people who just got their blood cholesterol tested.

Person	Concentration of Cholesterol in the Bloodstream	
	Low-Density Lipoprotein (LDL)	High-Density Lipoprotein (HDL)
A	3.62 mmol/L	0.90 mmol/L
B	2.33 mmol/L	1.94 mmol/L

a. Explain what the unit mmol/L means.

b. Explain which one of these people has the healthier cholesterol levels.

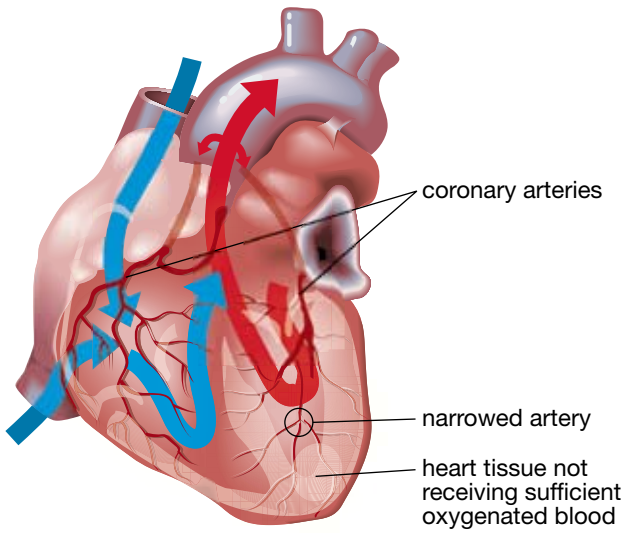
Atherosclerosis and Coronary Heart Disease

Atherosclerosis is the process in which deposits of cholesterol and fatty substances build up on the inside lining of an artery. This buildup results in a loss of elasticity or a hardening of the vessel. This condition may occur in any of the major body arteries.

When atherosclerosis affects coronary arteries that supply the heart with blood, complications result that affect the heart muscle's functioning. In this case, the disease is called **coronary heart disease**. Under these conditions the body's inability to supply the heart with sufficient oxygen leads to a buildup of toxic wastes. This causes a cramping pain called **angina**, which may begin behind the breastbone and then radiate out to the neck and arms.

- **atherosclerosis:** a hardening of the arteries due to the accumulation of fatty deposits
- **coronary heart disease:** a disease in which blood flow through the coronary arteries is restricted, possibly resulting in chest pain and/or a heart attack
- **angina:** chest pain caused by a narrowing of vessels that supply blood to the heart tissue

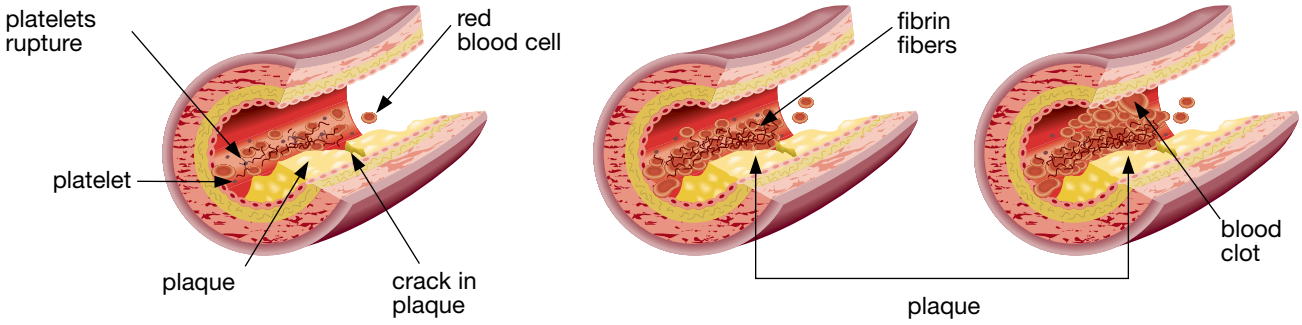
Coronary Heart Disease



Clots, Strokes, and Heart Attacks

If the plaque coating the artery becomes rough and cracked, platelets passing this area in the bloodstream can rupture and release chemicals that start the clotting process. Fibrinogen in the plasma is converted to thread-like fibrin, and red blood cells become trapped in the fibrin mesh to form a clot. The blood clot can completely block the flow of blood in an artery.

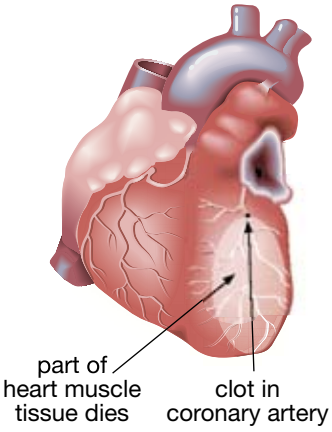
How Plaque in Arteries Promotes Blood Clotting



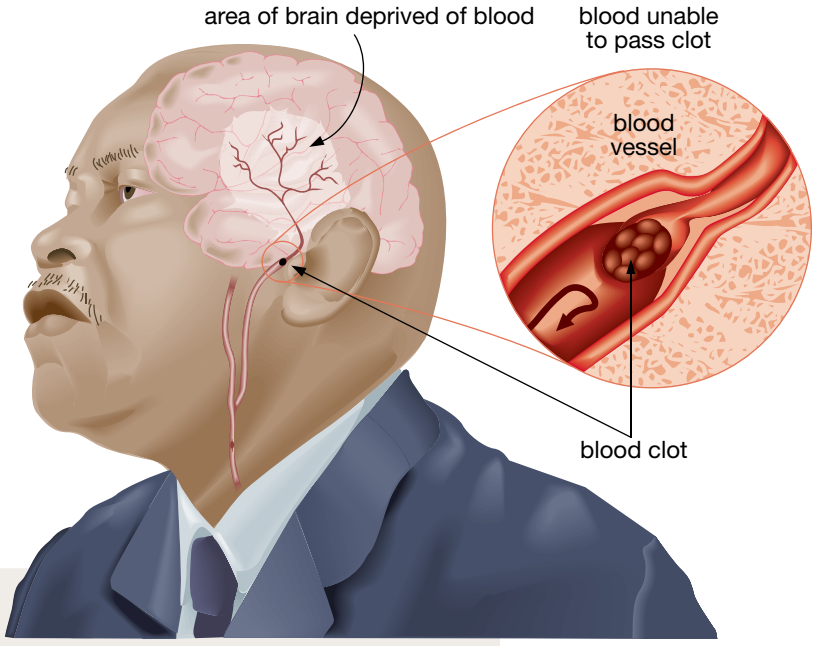
A blood clot in the coronary arteries, which supply the heart with blood, can damage the heart muscle and cause a **heart attack**. In this type of heart attack, cells of the heart muscle die because the clot prevents coronary arteries from supplying blood to those heart cells.

A blood clot in arteries supplying the brain with blood can cause a stroke. In a **stroke**, brain cells die. Memory loss, paralysis, or even death can result.

Heart Attack



Stroke



Practice

36. Explain the following statement.

> "Cardiovascular disease is a very broad term used to describe a collection of diseases and conditions. In some cases, one cardiovascular disease can cause another cardiovascular disease."

37. Earlier in this chapter you learned that a stronger heart is not a larger heart but a more elastic one. Elasticity—the ability to return to an initial form after being stretched—is a characteristic of a healthy heart. This same thinking also applies to healthy arteries.

a. Describe how the buildup of plaque reduces the elasticity of arteries.

b. Explain how the hardening and narrowing of arteries affects blood pressure.

38. Describe how the buildup of plaque on artery walls increases the likelihood of dangerous blood clots forming in the arteries.

- **heart attack:** the death of heart cells due to a blockage in the coronary arteries that supply oxygenated blood to the heart
- **stroke:** a sudden loss of brain function caused by an interruption in the blood flow to the brain

Fats and Cholesterol in the Bloodstream

Consider what you have learned so far in Lesson 1.4. Since the buildup of fat and cholesterol within arteries is one of the root causes of many cardiovascular diseases, you might be tempted to think that you should eat a low-fat, low-cholesterol diet. For many years this was considered sound advice for maintaining good health. However, if this were the whole story, how can you explain the fact that Inuit who eat their traditional diet have a very low incidence of cardiovascular disease, even though they obtain more than 50% of their calories from fat? Clearly there is more to this story.

Although food high in cholesterol can have an impact on the levels of cholesterol in your bloodstream, the typical person makes about 75% of the blood cholesterol in the liver, with only about 25% absorbed from food. Limiting the foods that contain cholesterol may not have a dramatic impact on lowering blood cholesterol levels. The real secret to lowering the levels of cholesterol in your bloodstream is to rethink the mix of fats in your diet.

The first distinction is between saturated fats and unsaturated fats. Recall from previous courses that saturated hydrocarbons contain only a single covalent bond between adjacent carbon atoms. Unsaturated hydrocarbons contain at least one double or triple covalent bond between carbon atoms. A saturated fat is made from long chains of fatty acids in which all the carbons are connected by single covalent bonds.

An unsaturated fat is made from long chains of fatty acids in which there is at least one double or triple bond. If there is only one double or triple bond, that fat is classified as monounsaturated. If there is more than one double or triple bond, it is classified as polyunsaturated. As shown in Figure A1.14, the presence of a double or triple bond in unsaturated fatty acids has a profound effect on the structure and, therefore, the properties of these molecules. Note that since the double bond occurs after the ninth carbon from the omega end, oleic acid is referred to as an omega-9 fatty acid.

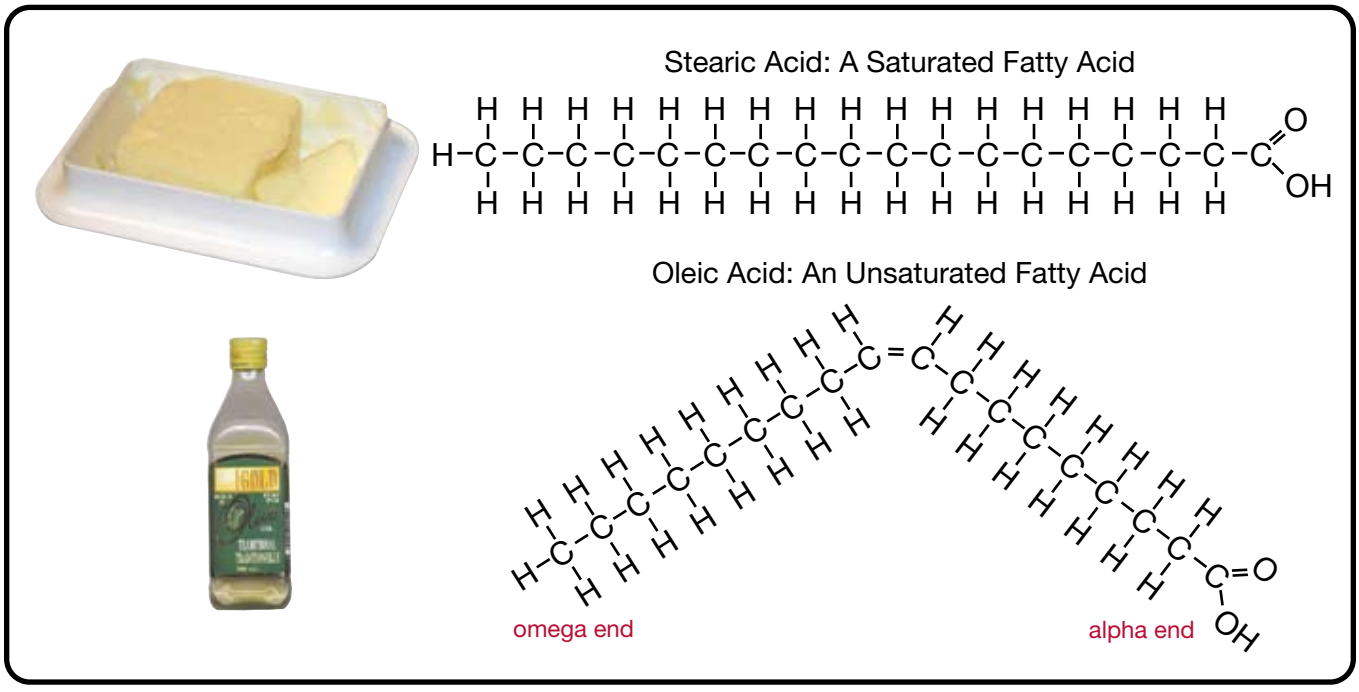
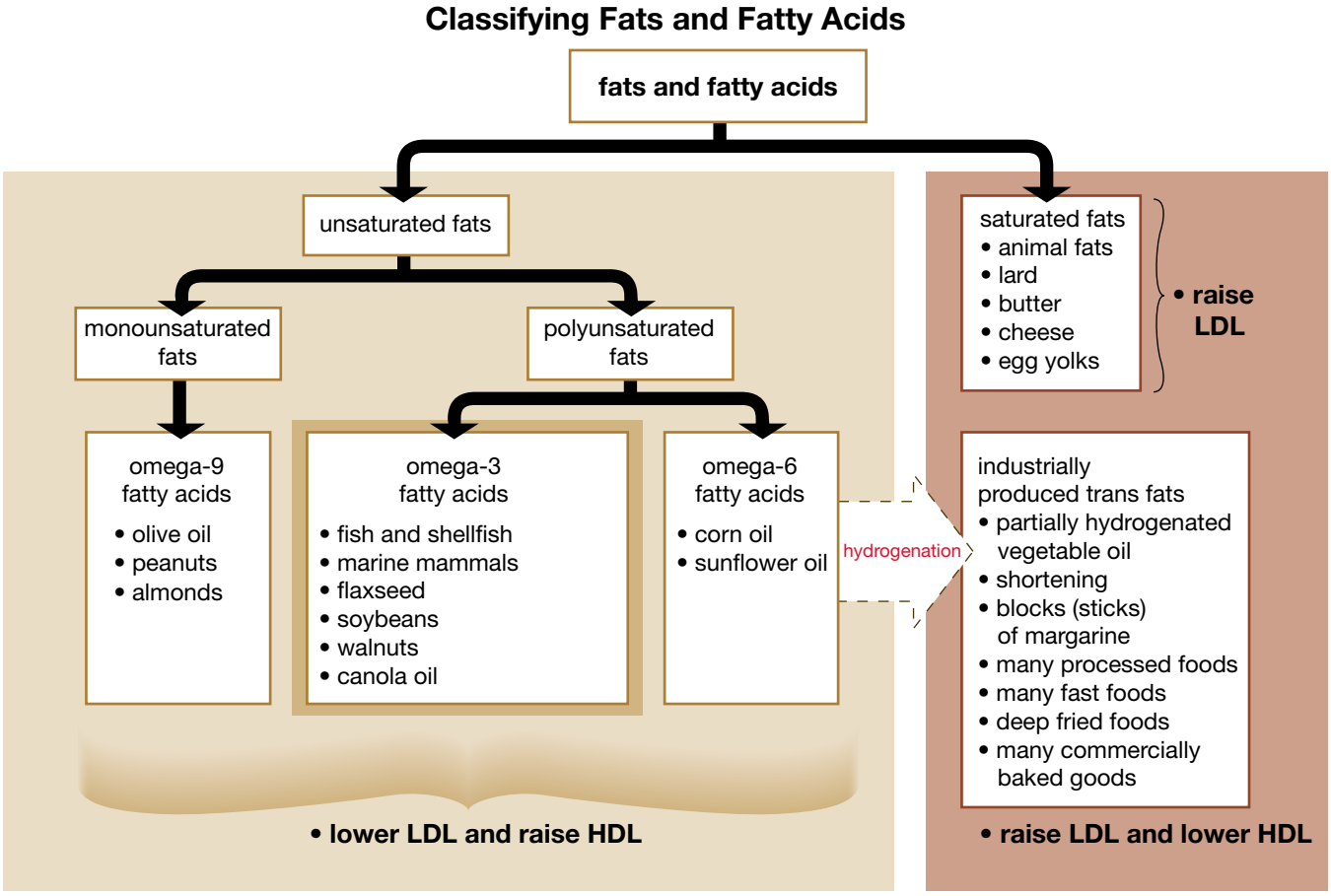


Figure A1.14: Butter is made of saturated fats, which contain saturated fatty acids. Olive oil is made of unsaturated fats, which contain unsaturated fatty acids.

So the types of fats and fatty acids you encounter at meal times can be categorized as either saturated or unsaturated. You may also encounter a third category of fats in the industrially produced trans fats. These compounds are created through an industrial process that involves bubbling hydrogen gas through hot vegetable oil under pressure in a special metal vat. In "Classifying Fats and Fatty Acids," trans fats are shown to be produced from vegetable oils, but they are organized under saturated fats because of the effects that trans fats have on cholesterol in the bloodstream.



Although this table contains lots of information, there are some important trends worth noting:

- The fats on the right have negative effects on blood cholesterol levels.
- The fats and fatty acids on the left have positive effects on blood cholesterol levels.
- The staple foods in the traditional Inuit diet—marine mammals and fish—are rich sources of omega-3 fatty acids. These substances have a positive effect on blood cholesterol levels.

Essential Fatty Acids

The fats listed on the left side of "Classifying Fats and Fatty Acids" are not only better than those on the right-hand side, but these substances are, in fact, essential for good health. You need these nutrients to form healthy cell membranes, to properly develop the brain and nervous system, and to produce hormone-like substances that regulate body functions (e.g., blood pressure).

essential fatty acid: a fatty acid that the body cannot synthesize itself and must be obtained from food

It is particularly important for you to eat foods with fats and oils that contain the **essential fatty acids** of omega-3 and omega-6. These substances cannot be produced from other substances within your body—you must obtain them from the foods you eat. Although most people have no trouble getting enough omega-6 fatty acids, many individuals find it challenging to obtain an adequate supply of omega-3 fatty acids.

Practice

39. Explain why limiting foods high in cholesterol can have only a limited effect on lowering blood cholesterol levels.

Use the information in "Classifying Fats and Fatty Acids" to answer questions 40 and 41.

40. In each part of this question you will be given a meal description. In each case, identify the dominant type of fat or fatty acid consumed and describe the likely effect on blood cholesterol levels.

- **a.** A day begins with a breakfast of bacon, eggs, and two pieces of buttered toast.
- **b.** A snack at a sporting event consists of a large order of deep-fried onion rings, a doughnut, and a large soft drink.
- **c.** A lunch at school consists of a tin of flaked tuna, along with a salad made up of fresh vegetables and homemade dressing created from olive oil and spices.

41. Suppose you decide to eliminate nearly all foods that contain fat. The only exception is that once in a while you treat yourself to a plate of fries with gravy from a local diner. Explain the negative impact of this eating pattern.

Heart Healthy Lifestyle

Males, individuals over 65 years of age, and people with a family history of cardiovascular disease are at the greatest risk for developing atherosclerosis and the potentially fatal circulatory problems that result from it. These risk factors cannot be controlled, but there are lifestyle choices that can affect your chances of getting atherosclerosis and related cardiovascular diseases.

If you routinely eat foods high in cholesterol, you may be negatively affecting your blood cholesterol levels. To maintain healthy cholesterol levels, keep in mind that the right mix of fats is actually more important than avoiding foods high in cholesterol. Nutritionists recommend trying to eliminate foods from your diet that contain industrially produced trans fats and add that you should replace foods containing saturated fats with foods that contain unsaturated fats.

People who have high blood cholesterol levels, who are overweight, who engage in little physical activity, who have high blood pressure, or who smoke (or even those exposed to high levels of second-hand tobacco smoke) are at a greater risk of developing circulatory diseases. Stress and excessive alcohol use can also strain the heart and blood vessels to increase the chances of developing a circulatory disorder. Making good lifestyle choices can help to ensure that you reduce the influence of factors that can harm your circulatory system.

DID YOU KNOW?

The number one cause of death in most developed countries is cardiovascular disease. Each year nearly 80 000 Canadians die from heart disease and strokes. This is the equivalent of the entire population of Red Deer.

Try This Activity

Analyzing Nutrition Fact Labels

Purpose

You will analyze nutrition fact labels to determine which foods are best for maintaining cardiovascular health.

Materials

You will need samples of nutrition fact labels from several brands of butter, margarine, and other spreads.

Procedure

step 1: Gather the nutrition fact labels from butter, margarine, and other types of spreads that you use most often in your home.

step 2: Create a chart to compare these products. The chart should include the product name, serving size, the food energy in calories, total fat, saturated fat, trans fats, cholesterol, fibre, and sodium.

Analysis

1. Rank the products in amount of cholesterol per gram of product and amount of saturated fat per gram of product. Identify which of the spreads would be best for someone trying to lower the blood cholesterol level.
2. A diet that includes lots of salty food causes sodium levels in the bloodstream to elevate. The body responds by adding more water to the bloodstream in an attempt to dilute the sodium concentration. Therefore, the volume of the blood increases.
   a. Explain why a diet that contains many salty foods leads to higher blood pressure levels.
   b. Identify which food items are best suited for someone diagnosed with high blood pressure.
3. Bile, mostly made of cholesterol, is a substance that helps to digest fat. Soluble fibre is found in foods like apples, brown rice, and beans. When soluble fibre passes through the digestive tract, it can help trap bile in the intestine—this allows bile to be excreted along with other wastes.
   a. Explain how soluble fibre helps to reduce blood cholesterol levels.
   b. Identify what food items contain significant amounts of fibre.
4. Other than differences in dietary content, list some factors that may influence consumer choice in terms of buying one of these products.

Omega-3 Fatty Acids and the Traditional Inuit Diet

Turn back to "Classifying Fats and Fatty Acids," which compared types of fats and fatty acids in terms of their effects on blood cholesterol levels. Note the location of fish and marine mammals on this table: these foods are rich sources of eicosapentaenoic acid (EPA), and docosahexaenoic acid (DHA). These particular omega-3 fatty acids can be derived only from marine sources—you cannot get them from plants or animals that live on land. These substances play a key role in allowing fish and marine mammals to survive in frigid environments. A good rule of thumb is that the colder the water, the greater the concentration of omega-3 fatty acids the fish and other animals can accumulate.

Figure A1.15: Salmon, along with mackerel and sardines, are coldwater fish and are all good sources of omega-3 fatty acids.

At the time this textbook was published, research on the benefits of a diet rich in omega-3 fatty acids from EPA and DHA marine sources was still underway. However, early findings indicate that these substances have a protective effect when it comes to cardiovascular diseases.

Possible Protective Effects of Omega-3 Fatty Acids on Cardiovascular Diseases

- helps prevent chaotic beating of the heart—the largest cause of death for people who have already suffered a heart attack
- reduces clotting of blood
- protects against progression of atherosclerosis by reducing harmful plaque
- reduces inflammation, especially associated with atherosclerosis in coronary arteries
- helps make arteries more supple and elastic
- causes slight reductions in blood pressure

It is important to realize that this research is still in its early stages and that the extent of these possible protective effects is still being investigated. How these protective mechanisms actually work is not yet thoroughly understood. Nevertheless, it does appear that the traditional ecological knowledge of the Inuit and medical science both support the same notion: a diet that contains foods like coldwater fish can have some positive effects on health.

It is also worth noting that the presence of omega-3 fatty acids from marine sources is not the only factor to consider when explaining the low incidence of cardiovascular diseases for Inuit who follow a traditional lifestyle. Genetics may also play a role. In addition, a lot of exercise accompanies the harvesting of food sources in the traditional Inuit diet. As you learned earlier in this chapter, people who include cardiovascular exercise or aerobic training as a part of their daily lives have stronger hearts that don't need to beat as often to circulate the same amount of blood.

Practice

42. Assume for the purposes of this question that research establishes there will be significant improvements to the cardiovascular health of the general population if people include marine sources of omega-3 fatty acids as an important part of their diets.

- **a.** A theme you encountered in previous science courses and will continue to explore in Science 30 is the notion that a technological solution to a problem often creates an unintended set of new problems. Identify what problem is addressed by having people increase the marine sources of omega-3 fatty acids in their diets.
- **b.** List some unintended new problems that could arise from this technological solution.
- **c.** How might some of the unintended problems identified in 42.b. be solved?

43. Refer to question 42. Some people argue that many of the difficulties created by technological fixes to problems stem from the fact that science can sometimes be characterized as "knowing more and more about less and less." In other words, science and technology can become intensely focused on the minute details of an extremely narrow field of study, and sometimes the "big picture" gets lost in the details.

- **a.** Review the definition for traditional ecological knowledge. How does this point of view help keep the "big picture" in focus?
- **b.** Comment on the possible benefits of integrating scientific research with traditional ecological knowledge.

44. Obtain a copy of the handout "Cardiovascular Disease Risk Questionnaire" from the Science 30 Textbook CD. Complete the questionnaire.

Add the completed questionnaire to your health file.

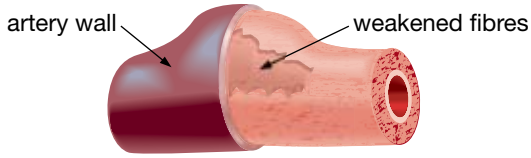
Other Cardiovascular Diseases

Not all cardiovascular diseases have their causes tightly linked to the effects of cholesterol and the types of fat present in a person's diet. Many cardiovascular diseases are due to injuries, conditions present at the time of birth, or a number of other factors.

Aneurysm

Have you ever turned on a garden hose and then bent the hose or pinched off the end of it so that the water couldn't come out? You might have noticed that pressure increased in the hose before the blockage. This extra pressure could cause the tough plastic surrounding the hose to weaken, bulge, and eventually tear. A stretched weakness in a blood vessel is called an **aneurysm**. An aneurysm can happen in any blood vessel, but it occurs most often in the aorta where blood pressure is highest. Sometimes an aneurysm is due to disease or injury, or it may be present from birth. Having an aneurysm is a dangerous condition because the stretched vessel wall is weak and could burst, causing internal bleeding. A ruptured aneurysm in the brain can cause a stroke or death. The development of an aneurysm can be caused by hypertension and atherosclerosis, but certain people have this condition at birth. An aneurysm is usually repaired by surgery, assuming the patient lives long enough to get surgery.

Aneurysm

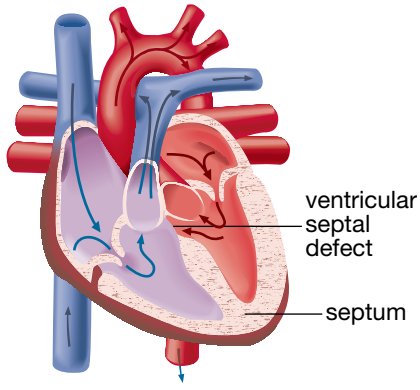


aneurysm: a widening or bulging of a blood vessel due to a weakening of the vessel wall

septal defect: a condition where the opening between the left and right halves of the heart fails to close before birth, causing excess blood to be pumped to the lungs

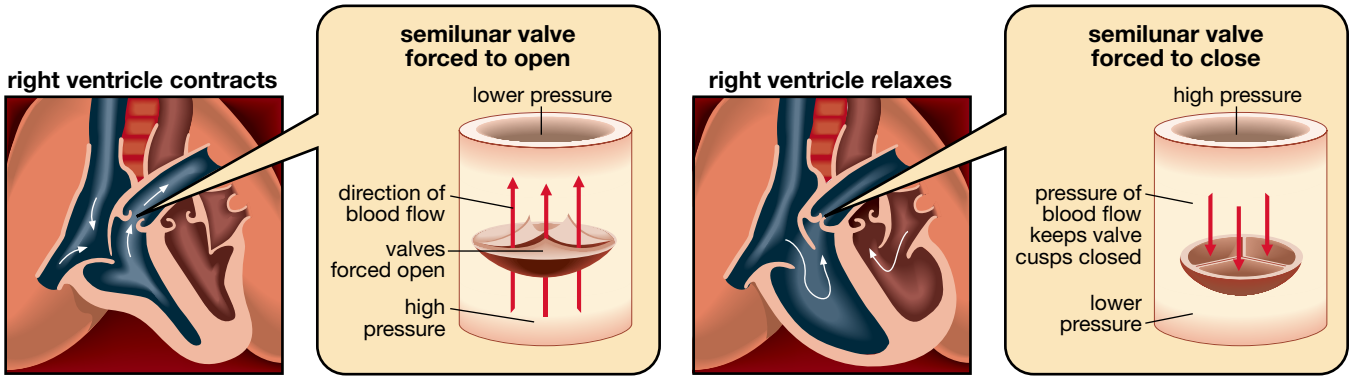
Septal Defect

When a fetus is developing in the womb, its blood doesn't need to go to its lungs to be oxygenated because the baby gets its oxygen from the mother's umbilical cord. Blood circulation in the fetal heart bypasses the lungs through an opening in the septum at the two atria—the top heart chambers. Before the baby is born, the hole between the two heart halves closes up. Occasionally, a baby will be born with a "hole in its heart." This **septal defect** causes some blood to flow into the right side of the heart, and excess blood is pumped to the lungs. The baby's septal defect usually closes up on its own, but larger holes often need surgery to be properly sealed.



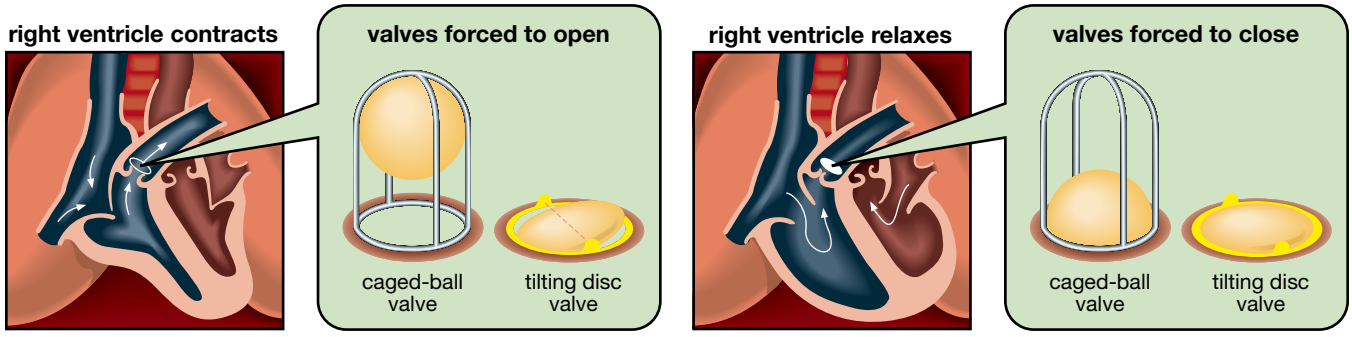
Valvular Heart Disease

Natural Heart Valves



Heart valves are used to control the direction of the flow of blood. If the valves don't close properly, blood can *backwash* against the direction of blood flow. If the valves are too narrow, insufficient blood is passed between the chambers and the heart must work much harder to circulate the required amount of blood.

Replacement Heart Valves



If a defective heart valve cannot be repaired, it is often replaced with an artificial valve or a valve made from human tissue or animal tissue. Artificial valves made from plastic or metal last for a long time but may cause blood clots. Recipients of artificial valves often have to take blood thinners for the rest of their lives. Valves made from human or animal tissue do not last as long as artificial replacements, but they pose less risk for the formation of blood clots.

1.4 Summary

Diseases or disorders of the heart and blood vessels that impair the functioning of the cardiovascular system are called cardiovascular diseases. Your risk for developing a cardiovascular disease may depend upon inherited genetic factors and lifestyle choices.

Atherosclerosis is a condition where a buildup of fatty substances, called plaque, coat the lining of arteries. This results in impaired circulation and heart pain (angina). Atherosclerosis can lead to the production of blood clots and even a vessel blockage. A blood clot in the coronary artery can cause a heart attack or, if it's located in the arteries leading to the brain, can cause a stroke.

Lifestyle factors that increase a person's risk of developing a circulatory disease include high blood cholesterol, high blood pressure, being overweight or inactive, smoking, high stress levels, and excessive alcohol use. Other cardiovascular diseases include aneurysms, valvular heart disease, and septal defect.

The traditional ecological knowledge of Inuit people has helped initiate scientific research. The fact that there is such a low incidence of cardiovascular disease among Inuit people who follow a traditional lifestyle—even though their diet is so high in fat—has acted as a trigger for current investigations. The importance of the types of fats and fatty acids in a person's diet is one of the major outcomes of this work.

1.4 Questions

Knowledge

1. Match each of the following terms with a description of its circulatory problem.

- plaque
- atherosclerosis
- angina
- heart attack
- stroke
- aneurysm
- septal defect

a. a chest pain during exertion due to constricted coronary arteries

b. a death of brain cells due to a blood clot in an artery supplying the brain with blood

c. a hole between the two halves of the heart that hasn't yet closed after birth

d. a hardening of the arteries due to a buildup of plaque in the vessel

e. a material with a rough, hard surface that forms on the inside of arteries due to the buildup of cholesterol and fatty substances

f. a condition that is caused by a blockage in the coronary arteries

g. a weakened bulge in a blood vessel that could rupture

Applying Concepts

2. A woman with a family history of heart and circulatory problems visits her doctor. List at least four things the doctor might ask about the patient's lifestyle, and describe changes the doctor might suggest to reduce the risk of the patient developing a circulatory disease.

3. Compare the analogy of a city's water delivery system to the human circulatory system. Explain what the following problems with a water delivery system can be compared to in the human circulatory system.

a. Something is stuck in one of the pipes and has caused some homes to lose water service.

b. The water pressure is so high that it is putting a strain on the pipes and causing them to leak.

c. A valve in the water pump is faulty.

Use the following information to answer questions 4 to 10. First Nations people who live in northern Alberta have acquired traditional ecological knowledge by living in the boreal forest for thousands of years. One element within this vast body of knowledge is that moose is a valuable source of food because it keeps people healthy. After scientific research, nutritionists have concluded that moose meat is a healthy food. The data in this table compares the nutritional value of raw moose meat to raw beef.

Nutrient (Serving Size)	Raw Moose Meat (100 g)	Raw Beef (100 g)
energy	427 kJ	1163 kJ
protein	22.24 g	17.48 g
total fat	0.74 g	22.55 g
• saturated fatty acids	0.22 g	9.16 g
• omega-6 fatty acids	0.14 g	0.57 g
• omega-3 fatty acids	0.03 g	0.23 g

4. Compare the total fat content to the serving size for both moose meat and beef. Express your answers as a percentage.

5. Account for the difference in food energy between the serving of moose meat and the beef serving.

6. Compare the saturated fat content to the serving size for both moose meat and beef. Express your answers as percentages.

7. Refer to your answers for question 6. Explain the significance of these numbers in terms of the effects on blood cholesterol levels.

8. Compare the omega-6 and the omega-3 fatty acid content to the total fat content for both moose meat and beef. Express your answers as percentages.

9. Refer to your answer to question 8. Explain the significance of these numbers in terms of the effects on blood cholesterol levels.

10. Refer to your answers for questions 4 to 9 to explain why moose meat is a good food choice for reducing the risk factors associated with cardiovascular diseases.

1.5 The Immune System

During medieval times in Europe, people worked very hard to farm and to tend their agricultural lands to produce enough food for their survival. Land was so valuable that wars often erupted over good farmland. Invading armies would attempt to take the land that people worked so hard to tend, so great fortresses were built for protection. If an army invaded, people from the surrounding countryside could move inside the castle to be protected by the thick castle walls. Any damage to the walls would quickly be patched by stonemasons. The castle could only be entered through guarded gates and across moats that could drown enemies.

Castle guards were posted along the walls to spot invaders, while other sentries patrolled and attacked any invaders who managed to sneak in. Different invading armies would use unique approaches and techniques to try to get into castles. After repelling the attack of an invading army, the defenders would be better able to respond to the same enemy if they tried to invade again.

Think of your body as if it were a medieval fortress. Different parts of your body act to maintain an ideal environment for the growth, health, and functioning of your cells. The organs in your body constantly work to provide the cells with a continuous supply of nutrients and to create a comfortable temperature and chemical environment. Unfortunately, the internal environment that the body works hard to create and maintain is also an ideal environment for the growth of disease-causing agents. These substances, often called germs or **pathogens**, are microscopic and act like foreign armies because they are constantly trying to invade the fortress of the human body.

pathogen: an agent, especially a virus or a bacterium, that causes disease

The human body has many ways to defend itself from pathogens:

- The skin covering the human body is a protective barrier, and it acts like the thick stone walls of a fortress. Skin is the first line of defence and prevents most disease organisms from getting inside the body.
- Because the sweat and oil secretions produced by the skin are acidic, bacterial growth is minimized.
- In Lesson 1.4 you learned about the role of platelets. These disks quickly create clots to repair damaged or broken skin to keep out foreign organisms. In a similar way, stonemasons repair damaged castle walls with replacement stones and mortar.
- The body has barriers that help defend these openings. For example, the nasal passage has hairs that work as filters, and mucous secretions trap most disease-causing organisms before they can reach the lungs. As well, the eyes are protected by an antiseptic liquid in your tears, and the stomach contains strong acid that kills most swallowed germs.
- The human body even has a defending army of white blood cells that act like soldiers—white blood cells attack pathogens that manage to break through the first line of defence to invade the body.

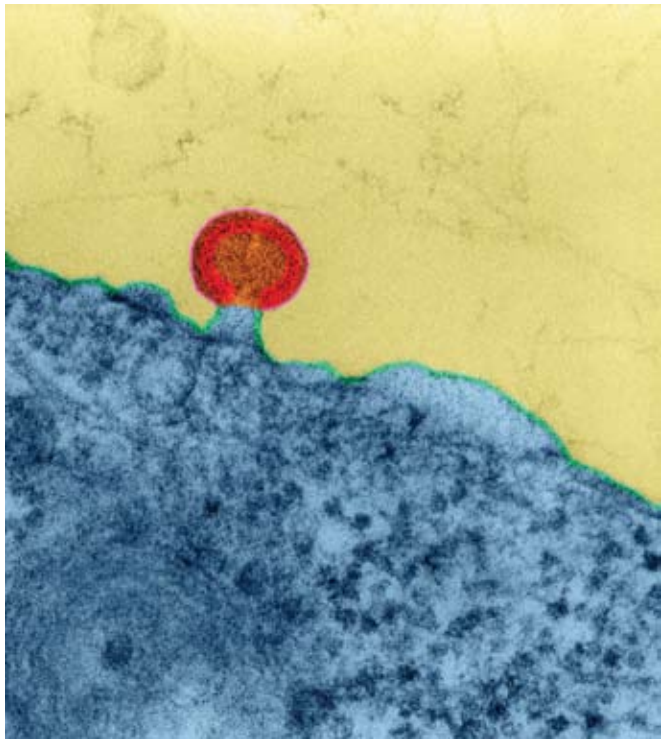


Figure A1.16: An HIV virus particle penetrates a cell membrane of human lymph tissue.

Practice

45. After reading the lesson introduction, copy and complete the following table that compares the roles and parts of a castle fortress to the parts of the immune system.

Part of Immune System	Role	Part of a Castle
		castle walls
cilia and mucous secretions		
	patch holes in protective barrier	
white blood cells		

Spreading Disease

If the body's natural defenses are not able to destroy or block a pathogen from entering it, the pathogen will begin to reproduce and spread. Even in the most sanitary living conditions, people regularly encounter microscopic substances that can get into their bodies, reproduce, and make them feel sick. These invading germ organisms can be passed along in several ways. When someone sneezes, coughs, or even talks, tiny droplets are expelled from the lungs. If this person's body has been infected by a disease-causing organism, like the influenza virus—which causes what is commonly referred to as the "flu"—or tuberculosis (TB), these expelled droplets will contain some of the pathogens. When other people breathe in these droplets, they can become infected by the pathogen.

DID YOU KNOW?

In some cases, bacteria and viruses can survive for months on the surfaces of everyday objects. When you touch an object, like the handle on an escalator in a shopping mall, you can transfer these pathogens to your hands and from there to your eyes, nose, or mouth, allowing pathogens to enter the body.

Regularly washing your hands is one of the best ways to avoid getting sick. All you need is soap and water. Ideally, you should rub your hands together for at least 15 s and scrub all surfaces including your wrists, under your fingernails, between your fingers, and the backs of your hands.

Food Poisoning

Pathogens can enter the body through the digestive system if contaminated food or water is ingested. The acid environment of the stomach is normally able to kill disease organisms. Food poisoning—an intense disturbance of the digestive tract—occurs when food is not cooked thoroughly, is improperly stored, or when the food is prepared in unsanitary conditions. For example, one type of food poisoning is caused by eating food contaminated with salmonella bacteria. Other instances of food poisoning can be seen in the "Common Types of Bacterial Food Poisoning" table.

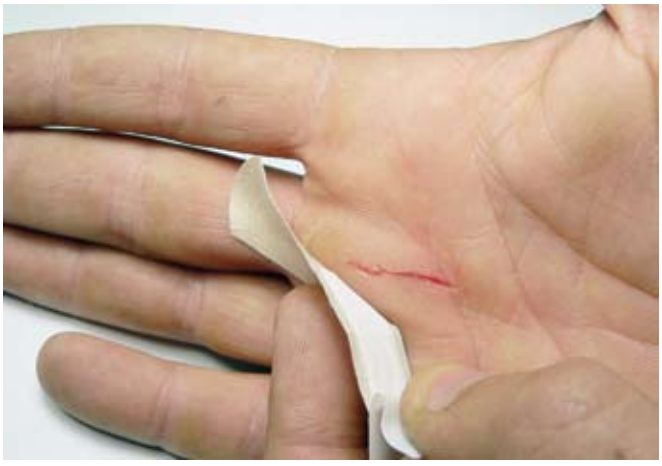
Cholera is a disease caused by a type of bacteria often found in dirty and untreated water. If the cholera bacteria are not killed by the low stomach pH, the bacteria can multiply in the intestine and infect the blood supply or release toxins that harm the body.

COMMON TYPES OF BACTERIAL FOOD POISONING

Bacterium	Habitat	Common Food Sources	Symptoms
salmonella	animal and human intestinal tracts	high protein foods like meat, poultry, fish, and eggs	diarrhea, vomiting, nausea, chills, and fever within 12 to 24 hours
clostridium botulinum (botulism)	soils, plants, marine sediments, and fish	improperly canned foods	blurred vision, respiratory distress, and possible death
listeria monocytogenes	soil, vegetation, and water—can survive for long periods in soil and plant materials	milk, soft cheeses, vegetables fertilized with manure	flu-like symptoms that mimic meningitis—elderly and babies most susceptible
E. coli (travellers' diarrhea)	feces of infected humans	meat and cheeses	diarrhea, abdominal cramps, no fever

Pathogens in the Bloodstream

HIV and the virus causing hepatitis C are examples of viruses transmitted through the bloodstream. When the skin is punctured or cut, pathogens can enter the bloodstream before platelets can seal up the breach. Cuts should be washed out and then covered with sterile bandages and dressings.



DID YOU KNOW?

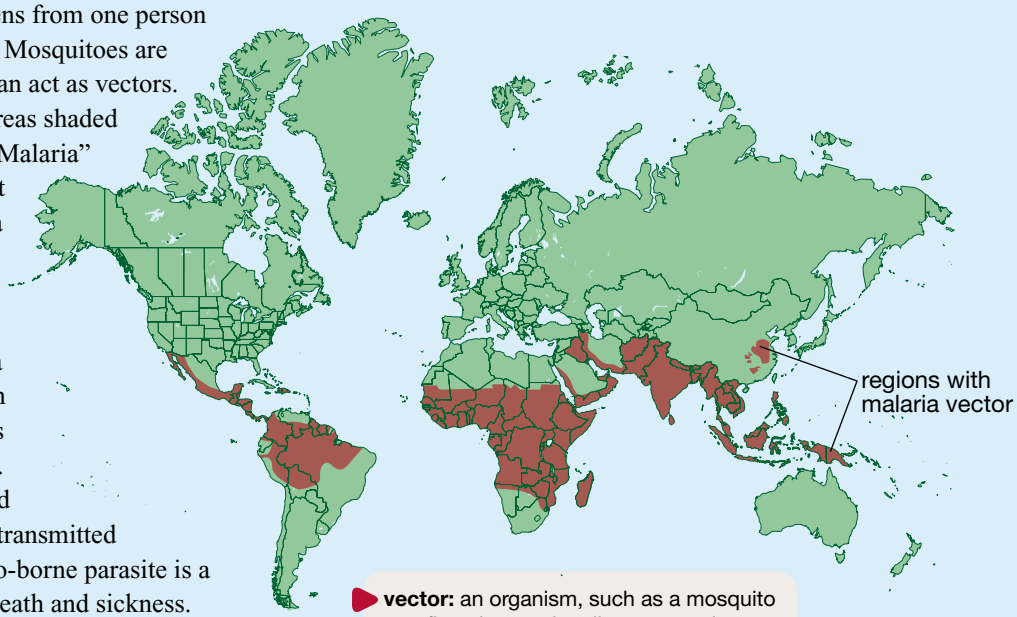
During the 1300s, an outbreak of the bubonic plague, or Black Death, occurred in Europe. This disease was carried by the fleas that initially lived on rats. After the rats died, the fleas passed the disease on to humans. In just five years, the Black Death killed about 25 million people, or the equivalent of one-quarter of Europe's population at that time.

There are still scattered cases of bubonic plague as the disease is often passed by flea bites from infected wild rodents—such as ground squirrels—to humans. If detected early, the disease is curable with modern medicines.

Vectors

Organisms that carry pathogens from one person to another are called **vectors**. Mosquitoes are examples of organisms that can act as vectors. In hot climates, such as the areas shaded brown on the "Prevalance of Malaria" map, mosquitoes can transmit the potentially deadly malaria parasite. When a mosquito punctures the skin to draw blood, it pumps some of its saliva into the bite. The saliva contains a chemical, called an anticoagulant, which prevents clotting while blood is drawn. The malaria parasite is carried in the mosquito saliva and is transmitted during the bite. This mosquito-borne parasite is a leading worldwide cause of death and sickness.

Prevalance of Malaria



vector: an organism, such as a mosquito or a flea, that carries disease-causing pathogens from one person to another

Try This Activity

Preventing Infection Poster

Design a poster that could be used to educate primary school students (grades 1 to 3) about the importance of good hygiene and practices that can help prevent infection by disease-causing pathogens. The poster should be colourful and attractive enough to appeal to young students.

Alternatively, your teacher could arrange for you to make a presentation to a primary school class on this topic or invite a primary school class to choose their favourite poster.

Joseph Lister (1827–1912)

During the nineteenth century, people who survived a successful medical operation often died due to infections that occurred during the operation. Most often, the infected wounds developed into gangrene or sepsis. Gangrene usually occurs in the extremities when cell tissues die because circulation has been lost in that area. A bacterial infection can cause a loss of circulation and result in gangrene. Sepsis is an illness that develops from a bloodstream infection by toxin-producing bacteria. At the time, there was not a complete understanding of how disease-causing agents were transmitted.

Joseph Lister was a British surgeon who studied Louis Pasteur's work on micro-organisms. Lister believed that hospitals needed to be clean and that he needed to kill unseen micro-organisms that were getting into wounds from the air. He began spraying a solution of carbolic acid onto wounds during operations and soaking dressings used for bandaging wounds in carbolic acid. This practice prevented the wounds from becoming septic. Lister's work with **antiseptics** reduced post-operation infections and saved many lives.

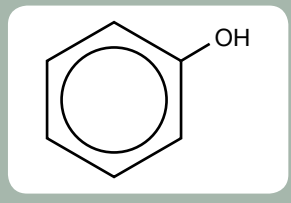
antiseptic: a solution or substance that prevents or inhibits the growth of micro-organisms

DID YOU KNOW?

The mouthwash called Listerine was developed in the late 1800s for use as an antiseptic for surgical procedures. Its inventors named the product after Joseph Lister, who pioneered antiseptic surgical procedures. It was soon discovered that the antiseptic solution was effective at killing mouth bacteria that cause bad breath and tooth decay. Listerine became popular and, in 1914, one of the first prescription drugs to be available over the counter.

Science Links

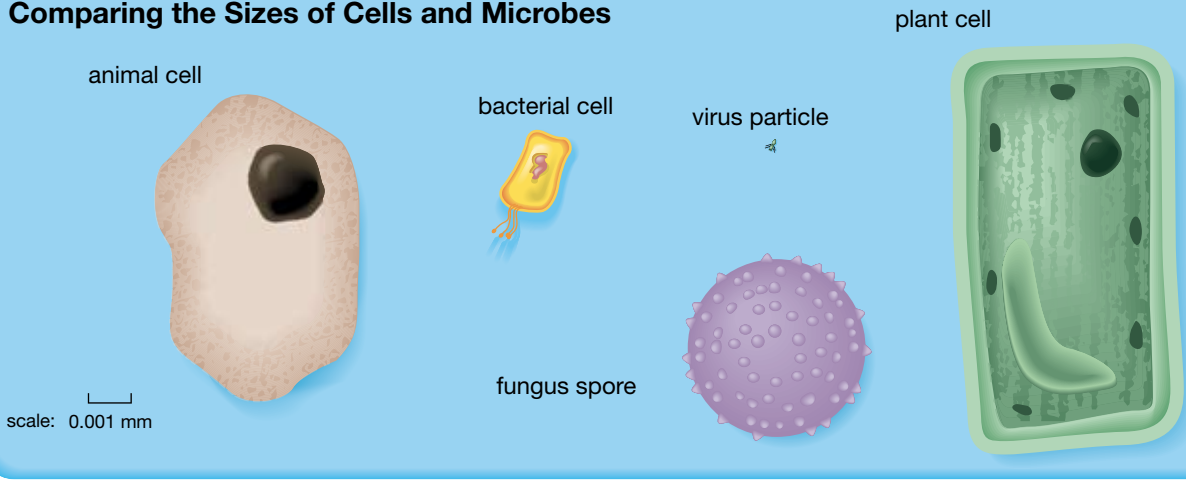
Carbolic acid is called *phenol* under the modern-day chemical naming system. Phenols are actually a group of compounds containing a ring of carbon atoms and an attached alcohol group. You will learn more about these chemical structures in Unit B. Phenols are still used as a component of commercial antiseptics.



Comparing Microscopic Pathogens

Micro-organisms live unseen all around. They are in the food and water that people ingest and in the air that they breathe. Most micro-organisms are harmless or even beneficial, such as those that live in your large intestine and play a role in digestion. If certain species infect a body or grow to large numbers, their negative effects cause the symptoms of disease.

Comparing the Sizes of Cells and Microbes



Protozoans

Malaria is caused by single-celled organisms called **protozoans**. Some protozoans live as parasites and require a host in which to reproduce. Because these protozoa exist with human cells, they are difficult to destroy without harming the host's cells. The protozoans that cause malaria are transmitted by a mosquito vector and infect human red blood cells.

protozoan: a group of microscopic, single-celled organisms that each have a nucleus

Many disease-causing protozoans can only divide within a host organism.

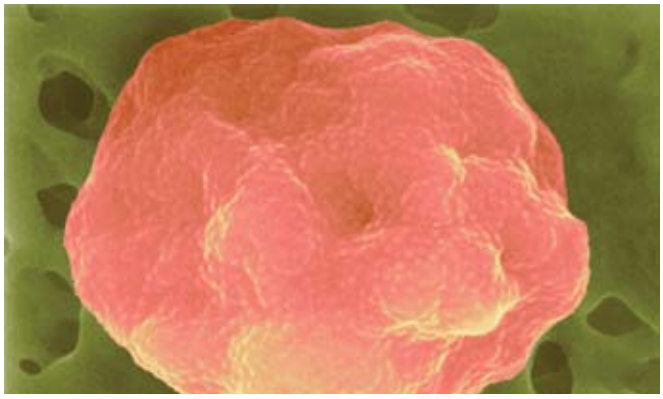
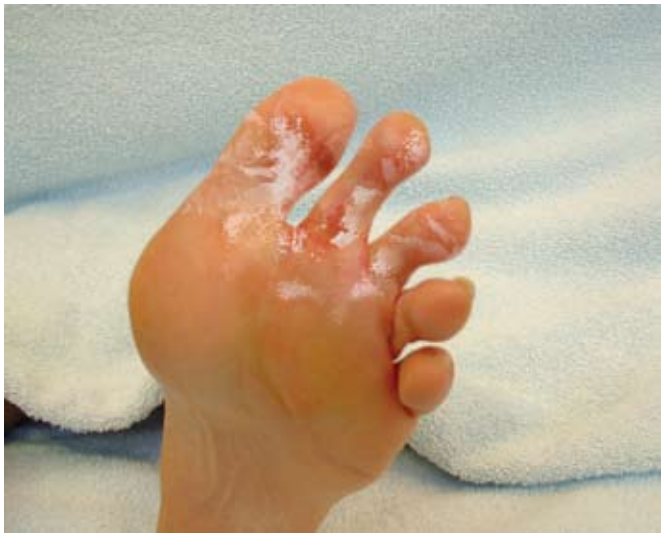


Figure A1.17: A red blood cell is infected with a malaria parasite.

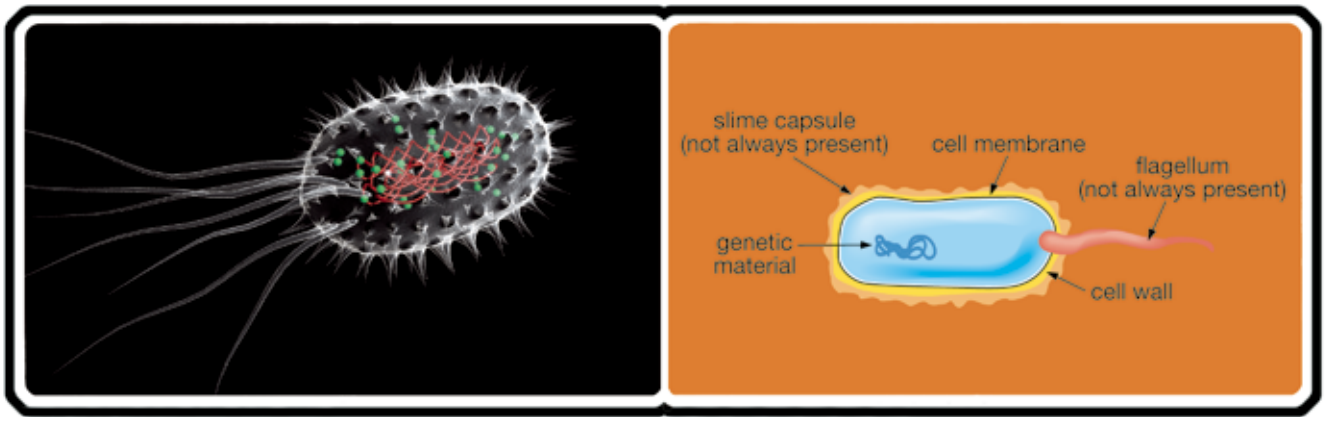
Fungi

Mold, mushrooms, and yeast are all examples of **fungi**. Most fungi live off the remains of dead or decaying organisms, but some are parasitic. Athlete's foot is an example of a fungal infection.

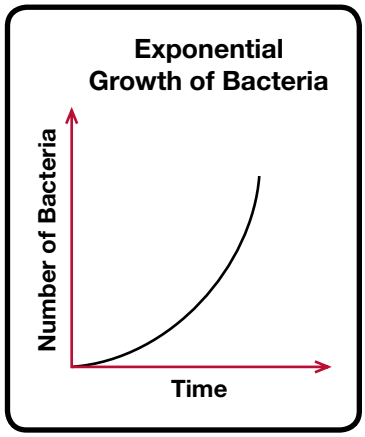
fungi: organisms that absorb food in solution directly through their cell walls and do not conduct photosynthesis; reproduction occurs through spores



Bacteria



Bacteria are small, single-celled organisms with a cell wall and cytoplasm. Unlike plant or animal cells, their genetic material is floating in cytoplasm and is not contained in a nucleus. Bacteria come in many different sizes and shapes including spiral-shaped, rod-shaped, or round. Some bacteria have a long whip-like tail—called a flagellum—or several flagella to help them move. Bacteria reproduce rapidly by simply splitting in two and can grow exponentially under ideal conditions. As disease-causing bacteria grow inside of you, their life processes damage your cells or they produce toxins that make you feel ill. **Antibiotics**, such as penicillin, are drugs that kill bacteria and, therefore, can be used to reduce or stop bacterial infections.



bacteria: microscopic, single-celled organisms that lack a membrane-bound nucleus and membrane-bound organelles; reproduction is chiefly by cell division to produce identical daughter cells

antibiotic: a drug that fights bacterial infections

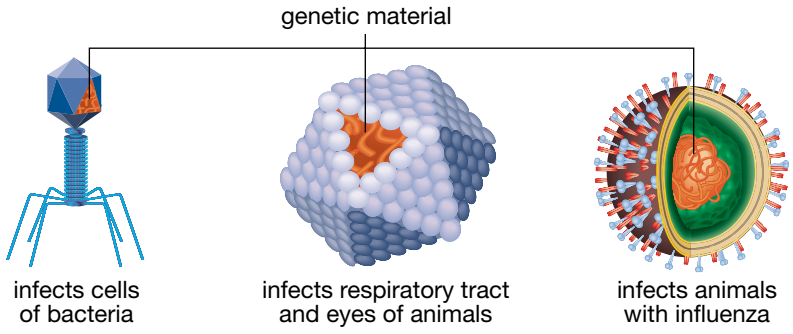
Viruses

Viruses are extremely tiny particles ($\frac{1}{100\,\text{th}}$ the size of a bacterium). Viruses do not grow, feed, or respire, so they are not considered to be cells. Scientists do not even consider them to be living organisms. They consist of a geometrically shaped protein coat and genetic material. Many viruses cause diseases. Viruses reproduce by infecting a host cell and injecting their genetic material into it, turning the host cell into a virus-making factory. Once new viruses are produced, the host cell ruptures and releases virus particles to infect more host cells. **Antiviral drugs** attempt to stop the infection of cells by viruses. These drugs also affect the development of new virus particles in the host cell.

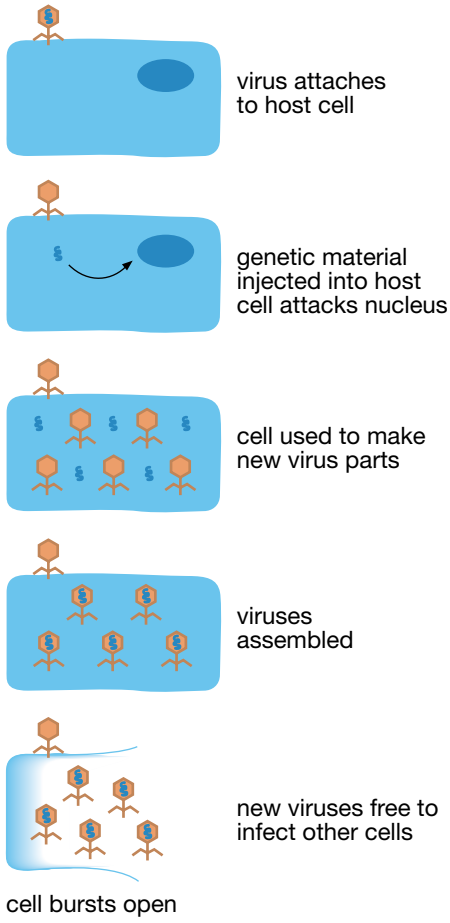
Bacteria and viruses are the most common types of disease-causing agents that can make people feel sick. The illness symptoms are due to tissue damage caused by these disease agents and by how bodies respond to this tissue damage.

- **virus:** a non-cellular particle consisting of a protein coat surrounding genetic material that multiplies only within the cells of a living organism
- **antiviral drug:** a type of medication that controls or cures an infection from a virus

Virus Particles



How a Virus Infects a Cell



Practice

46. Record and complete the following table in your notes.

MICROSCOPIC PATHOGENS

Type of Pathogen	Defining Characteristics	Example of a Disease Caused by This Type of Pathogen
protozoans		
fungi		
bacteria		
viruses		

47. In the late 1800s, scientists were struggling in their early attempts to isolate and identify viruses. Attempts to filter the particles responsible for viruses from infected plant fluids were unsuccessful, as were attempts to observe these particles in standard light microscopes. Identify the property of viruses that would account for these early difficulties.

Utilizing Technology

Informing the Public About an Infectious Disease

Purpose

You and your partners will develop a concise bulletin to inform the general public about an infectious disease. Your bulletin could take the form of a brochure, a poster, a multimedia presentation, or some other presentation to your class.

Science Skills

- ✓ Initiating and Planning
- ✓ Performing and Recording
- ✓ Analyzing and Interpreting
- ✓ Communication and Teamwork

Background Information

You have been employed by a public health clinic to produce an informative bulletin about an infectious disease. Health-care professionals wish to use the brochure you create to help patients become more informed about diseases. Your teacher may assign you a disease topic or you may choose a topic. Remember that your bulletin must be about an infectious disease like meningitis, strep throat, SARS, chicken pox, or hantavirus, and not about hereditary or environmental diseases and disorders like cancer, Down syndrome, or atherosclerosis. Check with your teacher before beginning if you are unsure about your chosen topic.

Materials

You will need to assemble the materials necessary for the bulletin format that your group plans to develop. You will also need access to the Internet, school library, and other resources to research your topic and to produce a product that summarizes the information in your own words.

Procedure

Read through the entire procedure. Then decide how you will divide up the tasks among group members.

step 1: The first task for your group is research. You may use the Internet and/or other resources to determine answers to the following questions:

- What background information should the public know about the disease you have chosen?
- What are the signs and symptoms of the disease?
- How is this disease transmitted?
- How can the disease be prevented and/or treated?
- Who is at risk for getting this disease?
- What do current statistics reveal about the number of people infected?

step 2: Plan how you can clearly communicate the answers to the questions in step 1 by using the format you have chosen. Your bulletin should be concise and effective.

step 3: Carry out the plan you devised in step 2 by preparing your bulletin about the disease you chose.

step 4: Share your bulletin with other students.

step 5: View the bulletins of other students.

Evaluation

1. Ask your classmates for feedback on the bulletin your group produced. How effectively did your group's bulletin address the six key questions from step 1? What aspects of your bulletin could be improved?
2. What did you learn from the bulletins prepared by other groups? If you completed this activity again, what would you do differently?

Immune Response

The immune system is like an internal army that fights off disease-causing organisms able to invade the body's first lines of defence.

The descriptions that follow match the graphic titled "Overview of Immune Response."

(1) The first event that initiates the process occurs when an invading pathogen breaks through the body's protective layer of skin and enters body tissues or the bloodstream. Fighting an infection begins with the detection of the disease-causing organism. Your internal army is composed of white blood cells that constantly check the identity of every substance encountered in the bloodstream to distinguish between the parts of your body and potentially harmful foreign parts. Each organism or virus displays unique chemical structures—usually proteins—on its surface. These structures are called **antigens**. Antigens on the outer surface of pathogens act like fingerprints to allow cells of the immune system to recognize these substances as potentially harmful foreign pathogens and to eliminate them from the body.

(2) It is the job of a type of white blood cell called a **macrophage** (literally meaning big eater) to patrol the bloodstream and eat dead cells, cellular debris, foreign cells, and molecules from outside the body. When a macrophage engulfs and destroys a disease-causing agent, it does not destroy the foreign antigen.

(3) Instead, it presents the invader's antigen on the surface of its cell membrane.

(4) Another group of white blood cells, called **T-cells**, mature in the thymus gland, which is a tiny structure behind the sternum. One type of T-cell—called a **helper T-cell**—binds to and recognizes antigens presented on the surface of a macrophage. The helper T-cell then serves to co-ordinate the remaining components of the immune system to respond to the invading pathogen.

(5) The helper T-cells can be thought of as the internal army's reconnaissance unit that provides vital information to co-ordinate an attack. Once the helper T-cells have recognized an antigen on a macrophage, they send out chemical messages to other groups of white blood cells.

(6) Helper T-cells alert the **B-cells**, which mature in the bone marrow. When the B-cells receive this chemical message from helper T-cells, they begin to multiply.

(7) Some of the B-cells produce proteins called **antibodies**. The antibodies produced by the stimulated B-cells are specific for each antigen presented on the macrophage.

(8) The antibodies attach to the antigens and sometimes stick to more than one invader by creating clumps of pathogens more easily engulfed by macrophages.

(9) The antibodies act like handcuffs to immobilize and tag the invaders for easier destruction by the macrophages.

(10) Helper T-cells also send chemical messengers to stimulate **killer T-cells**. Killer T-cells regularly patrol the body looking for cells that have changed due to mutation and could become cancerous. Since viruses replicate within body cells, the killer T-cells also look for cells that have been infected with viruses. The T-cells destroy these body cells by releasing proteins that create large holes in the membranes of the target cells.

(11) During the immune response, **memory B-cells and memory T-cells** are created and remain after the invading pathogen has been destroyed. The memory cells act like military intelligence archives by keeping a blueprint of the encountered invader's antigen to make the immune response quicker the next time that particular antigen enters the body.

(12) Once the invading organisms have been destroyed, another type of T-cell called the **suppressor T-cell** ends the battle by signalling the immune system to return to its pre-infection state.

antigen: a complex molecule on the surface of an invading pathogen that triggers an immune response

It is short for antibody generator.

macrophage: a type of white blood cell that engulfs dead cells, cellular debris, and foreign cells

It presents pathogenic antigens to T-cells in the immune response.

T-cell: a type of white blood cell that matures in the thymus gland

It recognizes and destroys invaders or releases chemical messengers to co-ordinate the immune response.

helper T-cell: a type of T-cell that co-ordinates the actions of other cells involved in the immune response

It sends chemical messages to activate the antibody producing B-cells and killer T-cells.

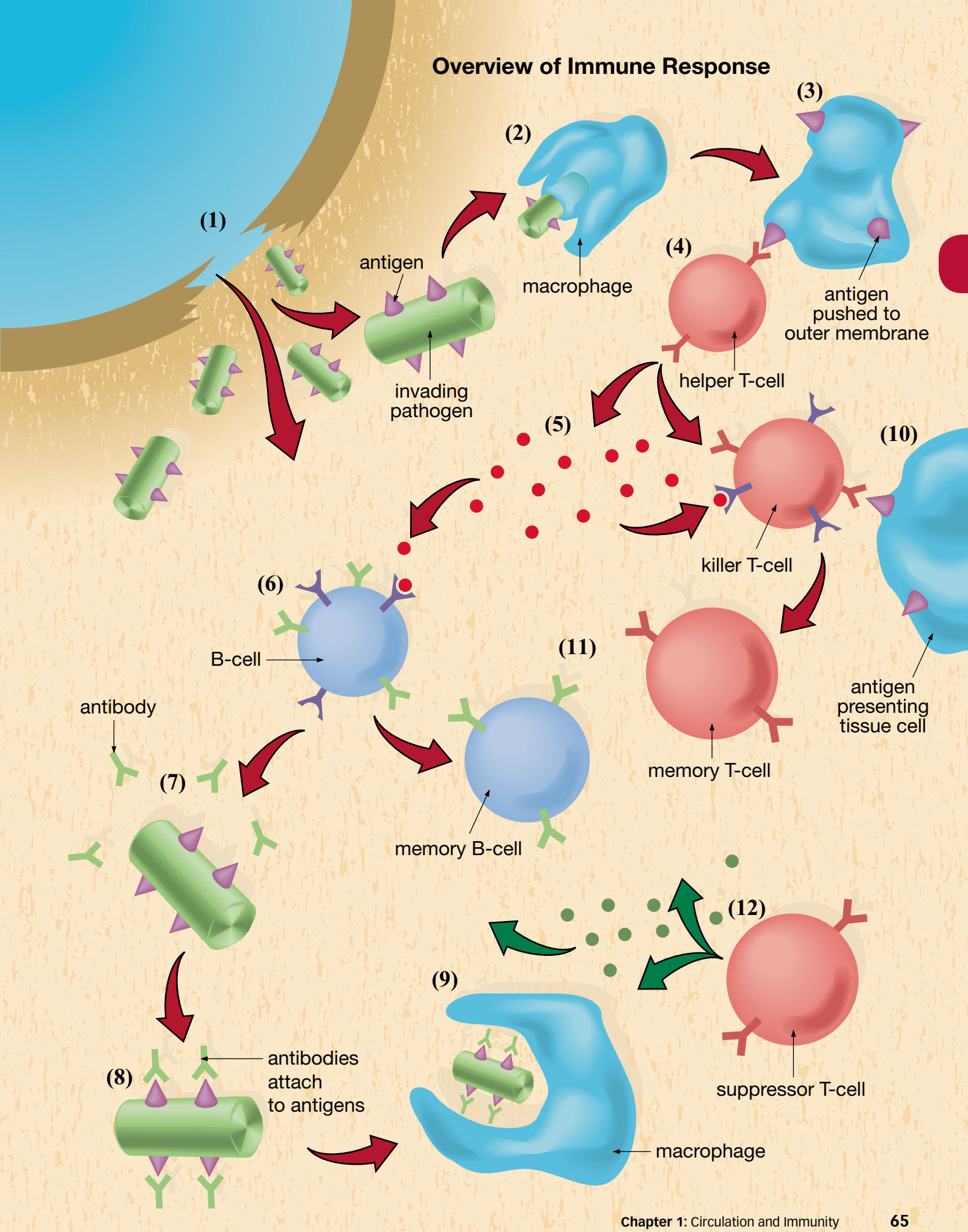
B-cell: a type of white blood cell that produces antibody molecules when stimulated by helper T-cells

antibody: a protein molecule produced by a B-cell designed to bind to a specific antigen to facilitate its destruction

killer T-cell: a type of T-cell that recognizes and destroys body cells by releasing proteins that create large holes in the target cell's membrane

memory B-cell and memory T-cell: specialized white blood cells that persist in the bloodstream to provide future immunity to invaders bearing a specific antigen

suppressor T-cell: a type of T-cell that sends chemical messengers to stop the immune response to an antigen


Overview of Immune Response
(1)
antigen
invading pathogen
(2)
macrophage
(3)
antigen pushed to outer membrane
(4)
helper T-cell
(5)
(6)
B-cell
antibody
(7)
(8)
antibodies attach to antigens
(9)
macrophage
(10)
killer T-cell
antigen presenting tissue cell
(11)
memory B-cell
memory T-cell
(12)
suppressor T-cell

Practice

48. Obtain a copy of the handout "Overview of Immune Response" from the Science 30 Textbook CD.

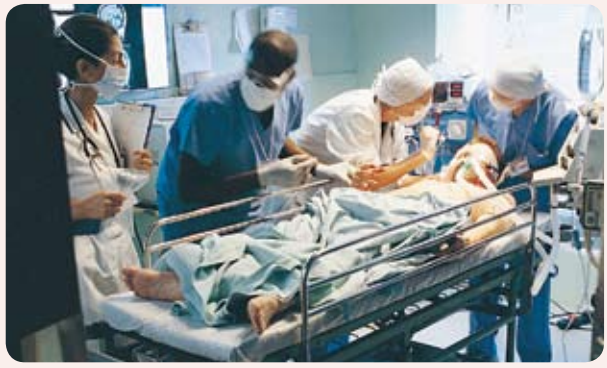
- **a.** Without looking at the labelled version of this illustration in the textbook, attempt to add the missing labels to this diagram.
- **b.** Once you have attempted 48.a., use the textbook illustration both to fill in labels that you were unable to complete and to check your work.

49. Obtain a copy of the handout "The Immune Response—Components and Roles" from the Science 30 Textbook CD.

- **a.** Without looking at information presented in the textbook, attempt to add the missing information to this table.
- **b.** Once you have attempted 49.a., use the information in the textbook both to complete and check your work.

DID YOU KNOW?

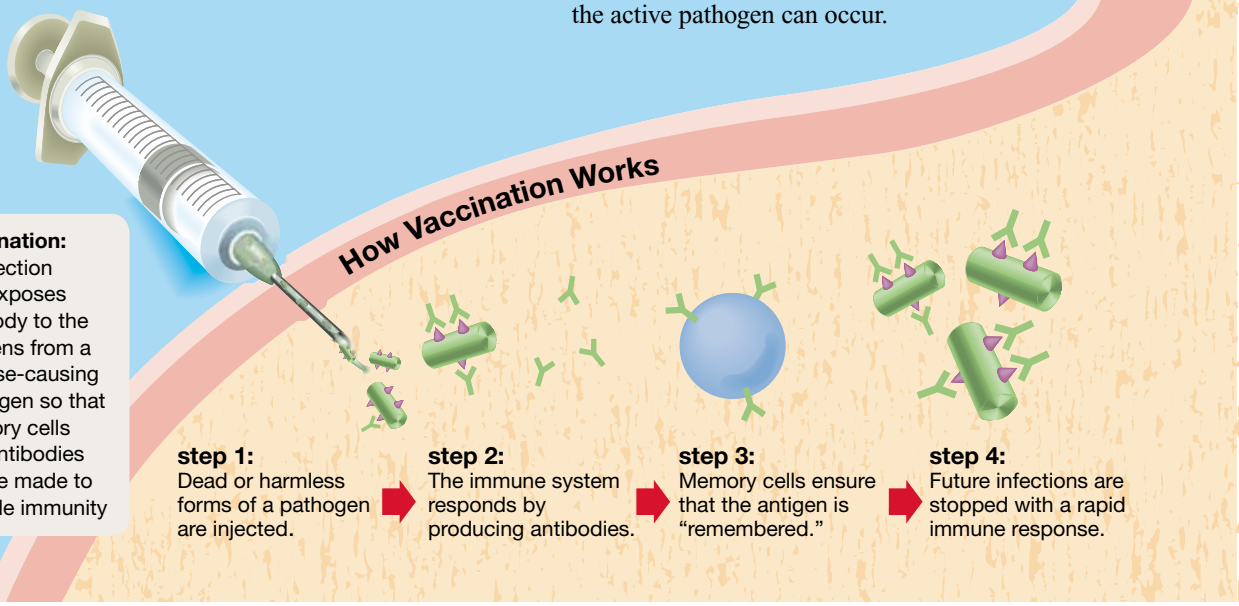
The presence of antigens on organs that are transplanted from one person to another also stimulates the recipient's immune system. The organ recipient's white blood cells often recognize the antigens on the donated cells and attack them by treating the organ like a foreign invader. To prevent a rejection of transplanted organs, transplant recipients may need to take drugs that suppress their immune systems for the rest of their lives.



Vaccinations

As long as the memory T-cells for a particular antigen remain in your body, they can provide long-term immunity to the diseases you have already encountered. The reason why people usually do not get chicken pox twice is because they retain memory T-cells for the chicken-pox virus. During a second exposure to an antigen for the chicken-pox virus, memory cells become rapidly activated, divide to form clones of themselves, and quickly produce large amounts of antibodies to act against the antigen. As a result, the invading organism is usually destroyed before it can bloom into a full-blown infection.

Immunity can be artificially developed by a **vaccination**. A vaccination—also called an immunization—involves the injection of an altered or weakened form of a disease-causing pathogen or an inactivated toxin into the body. An exposure to antigens allows the body to produce memory cells and antibodies against the disease. Because the substance used in a vaccination is either a weakened form or a killed form of the disease-causing agent, the risk of becoming sick from the disease is low. Booster shots are subsequent vaccinations of some of the material to ensure that memory cells exist so a quick and intense immune response to any future exposure to the active pathogen can occur.



In Canada, most people are vaccinated when they are babies against diseases such as measles, mumps, and rubella. People travelling outside of the country may get vaccinated against diseases they would normally not be exposed to in Canada, like yellow fever. Some people choose to be vaccinated each year against strains of the flu virus.

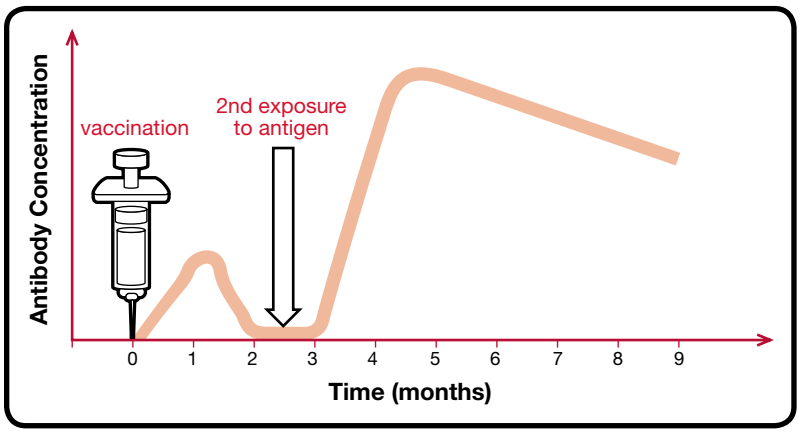


Figure A1.18: This graph shows how the concentration of antibodies is influenced by vaccination and a second exposure.

Practice

50. In your health file, list any vaccinations you have received. Alberta Health keeps a record of your vaccinations. You may need to ask a parent, a guardian, or a caregiver about vaccines you received as a baby.

Note: Some people have not received vaccinations for religious or other reasons.

Investigation

The Value of Mass Vaccinations: Weighing the Evidence

Background Information

There has recently been a debate over the benefit of administering vaccinations to masses of people within a population. Some individuals choose not to vaccinate their children or themselves. Many officials and professionals in the public health field maintain that the benefits of vaccinations still outweigh the risks.

Locate the following articles among the handouts on the Science 30 Textbook CD:

- "A Shot in the Dark"
- "Vaccine Myths and Why They Are Dangerous"

Purpose

You will consider two different opinions on the value of mass vaccinations. Then you will express your own opinion on this topic.

Procedure

step 1: Read through the "Analysis" and "Evaluation" questions to provide a focus for your reading.

step 2: Read each of the articles with the analysis questions in mind. Remember to be an active reader by using a highlighter or by taking notes as you read.

Science Skills

✓ Performing and Recording
✓ Analyzing and Interpreting

Analysis

A Shot in the Dark

1. List the negative effects of vaccination stated by interviewee Barbara Fisher.
2. State the reason Fisher gives to support a greater case for a connection between vaccinations and adverse reactions.
3. What personal experience has she had with vaccination?

Vaccine Myths and Why They Are Dangerous

4. List the negative effects of an unvaccinated population that are stated by author Dr. David Butler-Jones.
5. Describe his opinion on childhood vaccines being linked to seizure disorders, autism, and SIDS (Sudden Infant Death Syndrome).
6. What evidence does he provide to argue for a continued vaccination program?

Evaluation

7. Evaluate the two different sources for these articles. Do they seem like credible information providers?
8. Evaluate the two individuals who provide the information as either an interviewee or an author. Describe any bias or biases that they seem to present. Do you think that one individual is more credible than the other?
9. What is your stance on vaccinations? Against which diseases would you have your child vaccinated? Did your opinion change after reading the articles?

Utilizing Technology

Vaccinations for Travel

Background Information

Imagine that you have won a dream vacation! You can choose from a cruise down the Amazon River in Brazil, a visit to the famous Taj Mahal in India, an elephant ride in Thailand, or a safari tour in Kenya. Before going, you must find out what health precautions will be necessary for each of your destinations.

In this activity you will gather information from various sources of information, including the Internet, a public health centre, or a public health professional so you can answer each of the following questions.

1. List any vaccinations recommended or required before visiting one of these areas. Also, list the number of injections needed for a complete vaccination, the timeline for injections, the estimated length of immunity, and the cost, if any, for vaccination.
2. List any food or water-borne diseases to be concerned about in the location chosen, and describe precautions that will need to be taken.
3. List any vector-carried diseases—such as malaria—to be concerned about. Describe precautions that will need to be taken.

Science Skills

✓ Performing and Recording
✓ Analyzing and Interpreting

Edward Jenner

Smallpox is a potentially deadly virus that can kill as many as one-third of the people that it infects, and it leaves those who survive it disfigured with pockmarks. Smallpox has been a scourge for thousands of years. During European colonization and exploration, the population of many Indigenous peoples was decimated by their first exposure to smallpox in places such as North America, Central America, and Australia.

In the winter of 1781–82, fur traders on the North Saskatchewan River recorded going to First Nation encampments and finding the camps full of dead bodies. An explorer named Samuel Hearne estimated that 90% of First Nations people who traded at the nearby Hudson Bay Company post died of smallpox that winter.

Scientist Edward Jenner carried out a famous smallpox experiment in 1796, which led to the development of the first vaccine. He noticed that people, such as milkmaids, who worked closely with cattle were exposed to the much less deadly cowpox disease. People who had contracted cowpox were resistant to future smallpox infections. Jenner theorized that a human exposed to cowpox may develop some kind of immunity to smallpox—he tested this theory by taking pus from a milkmaid's cowpox sore and putting it into a cut on the arm of an eight-year-old boy named James Phipps, who was the son of Jenner's gardener.

After Phipps recovered from his cowpox infection, Jenner infected him with pus from a smallpox victim; but the boy did not become sick from the disease. This was the first example of an **inoculation**. Although people often use the words *inoculation* and *vaccination* interchangeably, inoculations introduce the antigens through a cut in the skin whereas vaccinations are performed through an injection of the antigens under the skin. The injection is done by a hollow needle.

We now understand that the antigen of the cowpox virus is similar to the antigen of the smallpox virus. By exposing the boy's body to cowpox, Jenner was able to rapidly produce antibodies to the smallpox virus, which prevented Phipps from becoming sick.

Due to aggressive vaccination programs designed by the World Health Organization (WHO), smallpox had been eradicated worldwide by 1979. As a result, most countries stopped vaccinating people for smallpox in the late 1970s.

inoculation: a process of producing immunity by introducing antigens of an infectious agent through a cut in the skin's surface

Practice

51. Evaluate Edward Jenner's investigative methods. Was it ethical to use James Phipps as a test subject?

52. Explain why Indigenous populations in North America, Central America, and Australia were particularly susceptible to the smallpox virus.

53. Samples of the smallpox virus are known to exist today in only a couple of laboratories.

a. Describe some concerns associated with keeping stocks of pathogenic, disease-causing agents.

b. Describe some benefits for keeping these stocks.

Autoimmune Diseases

Figure A1.19 shows the joints of a woman's hands that are greatly swollen with rheumatoid arthritis. This disabling and painful condition is an example of an **autoimmune disease**. Sometimes a person's immune system forms antibodies against his or her body's own tissues, treating them like the antigens of invading bacteria and viruses. The white blood cells act like a rebel army attacking specific body organs or causing a variety of illnesses. In this case, the person's white blood cells are attacking the bones and cartilage in the joints of her hands. The cause of autoimmune diseases is unknown, but scientists believe that the suppressor T-cells play a role in controlling the rebelling white blood cells. Studies indicate that autoimmune diseases are more common in women than they are in men. These diseases tend to occur later in life.

You have probably heard of multiple sclerosis (MS) and diabetes mellitus (type 1 diabetes). Both are examples of autoimmune diseases. With MS, white blood cells attack parts of the nervous system. In the case of type 1 diabetes, the body mistakenly manufactures antibodies directed against the pancreas. The result is that the pancreas is unable to make insulin, which is a hormone that helps regulate the concentration of glucose in the bloodstream.

autoimmune disease: a disorder in which the immune system produces antibodies against the body's own cells

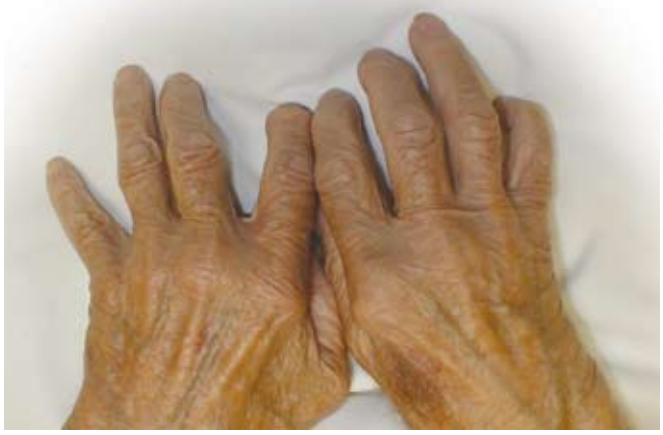


Figure A1.19: Rheumatoid arthritis can be a very debilitating disease.

1.5 Summary

The human body's internal environment is ideal for the growth of many disease-causing organisms, so the body must have defenses to protect itself from disease. The skin, cilia, and secretions—such as stomach acid, tears, and mucus—act as the first line of defence against potential invading organisms. White blood cells act as a defending army to identify and destroy any disease-causing organisms that make it through the skin and secretion barrier.

Disease-causing organisms can be passed on in several ways. These include being spread through droplets in the air, by eating or drinking contaminated food or water, via cuts in the skin, or through vectors such as mosquitoes. Disease-causing organisms, including protozoans, fungi, bacteria, and viruses, all have distinctive antigens on their surfaces that allow the white blood cells to recognize them as foreign invaders.

The white blood cells of the immune system are specialized for specific functions. Macrophages ingest disease-causing organisms displaying antigens from the destroyed invader. Helper T-cells recognize antigens and co-ordinate the attack against the invaders. B-cells produce antibodies that bind to the disease-causing organism's antigens. Killer T-cells destroy virus-infected body cells. Suppressor T-cells end the immune response. Memory T-cells and memory B-cells remain to provide a faster response to subsequent encounters with the antigen.

Vaccinations are a way of artificially exposing someone to an antigen of a disease-causing organism so that the body can produce memory cells and antibodies to create a greater immunity to exposure from the antigen.

1.5 Questions

Knowledge

1. Explain how each of the following disease-causing organisms overcomes the body's natural defenses to enter the body.
   - **a.** malaria
   - **b.** hepatitis C
   - **c.** tuberculosis
   - **d.** salmonella
2. Explain how an autoimmune disease differs from an infectious disease.
3. Create a table comparing the similarities and differences between bacteria and viruses.
4. Locate the applet "Battles in the Bloodstream" on the Science 30 Textbook CD. Watch the applet to confirm your understanding of how the immune system responds to a microscopic pathogen. Turn the sound off. Then watch the applet again. As you watch it, supply a new audiotrack by providing a description of what is occurring. Be sure to include the following key words in your spoken commentary: macrophage, antigens, helper T-cells, B-cells, antibodies, killer T-cells, suppressor T-cells, memory B-cells, and memory T-cells.

Applying Concepts

5. Explain how the following problems impair the body's ability to fight against disease-causing organisms.
   - **a.** A person with hemophilia has blood that does not clot properly.
   - **b.** The skin is badly damaged so that tissues are exposed.
   - **c.** Someone who has HIV has many helper T-cells destroyed by the virus.
6. Explain how the following methods can be used to assist the body's ability to fight against disease-causing organisms.
   - **a.** vaccinations given at birth
   - **b.** antibiotics prescribed when you have an infection
   - **c.** antiseptics used during operations
7. In a group, act out for the class the body's immune response to an invading bacteria or virus. Each group member will be assigned the role of one immune response component (disease-causing organism, macrophage, helper T-cell, B-cell, killer T-cell, suppressor T-cell, or the memory T-cell). Small groups may need to have members perform the role of more than one component. Before performing for the class, your group may want to spend some time developing a script, finding costumes for different roles, and rehearsing.

Chapter 1 Summary

In this chapter you have examined the structure and function of the circulatory system. You have seen how the heart functions to pump blood through the body, how the vessels of the circulatory system are specialized to carry blood around the body, and how blood functions as a medium to transport substances and to protect against disease-causing pathogens. You have also seen how problems that affect the circulatory system impair its functioning, and you have examined some of the technologies used to treat these problems.

Most diseases that make us feel sick are caused by lifestyle choices or by the invasion of disease-causing organisms. In your health file, you have examined and recorded information about your health and risk factors. In Chapter 2 you will look at the mechanisms of inheritance and learn how certain diseases can arise from inherited traits rather than from the environment. You will also examine the ethics of using genetic technologies.

Summarize Your Learning

In this chapter you have learned a number of new biological terms, processes, and theories. It will be much easier for you to recall and apply the information you have learned if you organize it into patterns.

Since the patterns have to be meaningful to you, there are some options about how you can create this summary. Each of the following options is described in "Summarize Your Learning Activities" in the Reference section. Choose one of these options to create a summary of the key concepts and important terms in Chapter 1.

Option 1: Draw a concept map or a web diagram.	**Option 2:** Create a point-form summary.	**Option 3:** Write a story using key terms and concepts.	**Option 4:** Create a colourful poster.	**Option 5:** Build a model.	**Option 6:** Write a script for a skit (a mock news report).

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Chapter 1 Review Questions

Knowledge

1. List the four main functions of the human circulatory system.
2. Copy and complete the following table comparing the chambers of the mammalian heart.

Heart Chamber	Location	Type of Blood Found in Chamber	Function
right atrium	top right	deoxygenated	receives blood from body from vena cava
right ventricle			
left atrium			
left ventricle			

3. Describe three ways in which arteries and veins differ.
4. List the four main components of blood. Rank these components by their relative proportion in a blood sample from the largest proportion to the smallest proportion.
5. State which of the four major blood components is responsible for initiating the clotting process.
6. Define *cardiovascular disease*.
7. Distinguish between a heart attack and a stroke.
8. List four ways by which disease-causing pathogens can enter the body.
9. Define *vaccination*.
10. Obtain a printed copy of the handout titled "Overview of Immune Response" from the Science 30 Textbook CD.

    a. Use a pair of scissors to cut out all 13 images.
    b. Without looking at the textbook, place these images in the correct sequence.
    c. Check your answer to question 10.b. with the information presented in this chapter. Make the necessary adjustments or corrections and then use a glue stick or transparent tape to attach the images to a piece of paper in the correct sequence.

Applying Concepts

11. From the following data, carefully examine the relationship between the heart rate and the mass of an organism.

RESTING HEART RATE VERSUS MASS

Organism	Mass (g)	Resting Heart Rate (beats/min)
mouse	25	670
rat	200	420
guinea pig	300	300
rabbit	2000	205
small dog	5000	120
large dog	30 000	85
human	70 000	72
horse	450 000	38
African elephant	6 000 000	30

a. Observe trends from this data. Write a statement that describes how heart rate is affected by the size of an organism.

b. Estimate the heart rate of a 3-kg cat from these data patterns.

c. *Tyrannosaurus rex* was a ferocious carnivore that lived from about 85 to 65 million years ago. Paleontologists estimate that T.rex had a mass of up to 7000 kg. Estimate the resting heart rate of T.rex by extending the trends in this data.

d. The extinct dinosaur *Apatosaurus* (also called *Brontosaurus*) belonged to the long-necked family of sauropods. This family included the largest land animals to ever live. Estimate the heart rate of *Apatosaurus*, which had a mass of 27 metric tons (27 000 kg) by extending the trends in this data.

e. What problem does extending the trends in the data pose for paleontologists who are studying the circulatory systems of large extinct dinosaurs?

Use the following information to answer questions 12 to 14.

In Lesson 1.1 you learned how the parts of the heart work together as a system. A great way to consolidate all that you have learned about the heart is to build a three-dimensional model of this amazing pump.

To build the model, you will need the following:

- modelling materials such as Femo, Plasticine, or playdough
- a piece of stiff cardboard to act as a base for the model
- a copy of the handout titled "The Human Heart—Labelled" from the Science 30 Textbook CD

Note: You can make your own playdough by using one of the recipes on the "Playdough Recipes" handout on the Science 30 Textbook CD.

Design Criteria for the Heart Model

These models are designed to be three-dimensional representations of the handout titled "The Human Heart—Labelled." All the key structures on the heart diagram are to be included in your model. Even though the model will not include labels, if someone points to a heart structure on your model, you should be able to answer the following focusing questions:

- What is the name of this structure?
- Where would blood in this part of the heart go next?
- Does this part contain oxygen-rich blood or oxygen-poor blood?

12. Build your three-dimensional model of the heart according to the design criteria. Make sure all the parts connect so that if the parts were hollow, blood could flow through the heart. Building the model on a piece of stiff cardboard will make it easier to clean up and to move the finished product.

13. When your model is complete, show your work to other students. Ask the other students their opinions about what are the most effective aspects of your model as well as how it could be improved. Provide similar feedback to other students who completed this activity.

14. Use the feedback you received from your classmates as well as what you observed in the other models to answer the following questions.

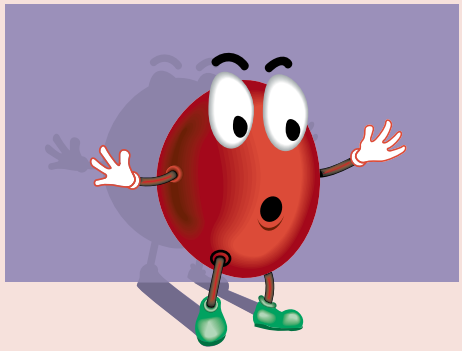
a. What are the strong points of your model?

b. How could your model be improved?

Use the following information to answer question 15.

You have been asked to volunteer to help children in elementary school learn about the circulatory system. Your task is to create a comic strip to accompany the following description in an elementary school resource.

Robbie the Red Blood Cell is in the vena cavae on his way back from the brain where he just delivered some fresh oxygen to a brain cell and grabbed some waste carbon dioxide. While in the brain, Robbie found out that his friend Tina Toe Cell is in desperate need of oxygen.



15. Create a colourful and attractive comic strip with a series of six to eight sequential panels that explain what happens to Robbie as he moves through the circulatory system from the vena cavae to the toe and back again. You should aim to design a comic strip that could teach about the pathway of blood in the human body to someone who has never studied the circulatory system.

16. The bodies of athletes who compete in endurance events—such as marathon runs, cross-country skiing, or bike races—require a huge amount of oxygen during the competition. Some endurance athletes have tried to improve their performance by removing their own blood, centrifuging it to isolate the red blood cells, storing it while the body replaces the lost blood, and then injecting it back into their own body right before the race. This process of "blood doping" has been banned by the International Cyclist Union (UCI) and also by the International Olympic Committee (IOC).

a. Why would injecting more red blood cells into their bodies create an advantage for athletes?

b. Explain why it is more difficult to prove that athletes are using blood doping rather than taking performance-enhancing drugs.

c. Predict the effects of blood doping on the athlete's blood pressure.

d. List some possible negative health effects of the practice of blood doping.

e. Athletes often train at high altitudes before a competition. The thin air at these altitudes stimulates red blood cell production. Some athletes feel that the practice of blood doping before a competition is no different than training at high altitudes. Evaluate this specific argument.

17. Design an Experiment

A new over-the-counter weight-loss drug has just been released onto the market. Most users of the product are reporting fantastic weight-loss results. However, some clients are reporting an increase in their heart rates as a side effect. The drug company claims that there is no connection between these reported heart effects and the proper use of their product. A regular user of the product recently died of a heart attack. There is now public concern about the drug's use.

Your job is to design an experiment that safely tests whether there is a correlation between using the drug and risking an increased or irregular heartbeat that could result in a heart attack. A correlation is an assessment of how strongly two variables are related. If one variable changes and the other variable changes with it, there is said to be a correlation.

Provide a description of an investigation you would carry out, what materials you would use, and how you would ensure that the experiment was done safely. Your experimental design should clearly identify the manipulated and responding variables as well as listing at least three controlled variables. It should also take into account ethical and safety considerations.

18. Whales and seals are mammals well adapted for diving. For example, the Weddell seal is able to remain underwater for over an hour without surfacing to breathe. List some possible adaptations of a diving mammal's circulatory system that would allow it to remain below the water for so long.

19. A doctor looks at three patient files containing information from lab tests and lifestyle data. Note that μL is a microlitre.

Health File Information	Patient 1	Patient 2	Patient 3
Cholesterol Level	200 mg/dL	280 mg/dL	150 mg/dL
Activity Level	moderate regular weekly exercise	little or no weekly exercise	intense physical training
Resting Heart Rate	72 beats per minute	81 beats per minute	50 beats per minute
Smoker?	occasionally	yes	no
Blood Pressure	120/80 mmHg	147/95 mmHg	120/80 mmHg
White Blood Cell Count	14 000 per μL	6500 per μL	5000 per μL

a. Explain which patient the doctor would be most concerned about in terms of circulatory health? What lifestyle changes or future precautions might the doctor recommend to the high-risk patient?

b. Which patient most likely has an infection?

c. State the likely reason why the resting heart rate of Patient 3 is significantly lower than the other two patients.

Use the following information to answer questions 20 to 24.

Investigating Primary Literature: Predicting Sudden Death

Cardiovascular disease is the number one cause of death in Canada, with nearly 80 000 annual victims. For almost half of these people, death occurs within minutes of the first symptoms of a heart attack. In many cases the first heart attack pains were the first indication that this person had poor cardiovascular health—but by then it was too late! Clearly, if there is a way to predict whether an apparently healthy person is at risk of sudden death from a heart attack, preventative measures can be taken and lives can be saved.

Researchers from France and Italy analyzed data over 23 years from men working around Paris in the French civil service. The researchers claim that they have found a practical way to predict which members of a healthy population of men, who have no previous history of cardiovascular disease, may be susceptible to sudden death from heart attack.

This research is described in the article "Heart-Rate Profile During Exercise as a Predictor of Sudden Death." This article is found as a handout on the Science 30 Textbook CD. Since this article was published in the *New England Journal of Medicine*, it can be a challenging piece to read because it was written for physicians and medical science researchers. Nevertheless, there is value in reading primary literature because you can learn about scientific discoveries as they are reported by those people who actually did the research.

20. Obtain a copy of the handout "Heart-Rate Profile During Exercise as a Predictor of Sudden Death" from the Science 30 Textbook CD.

- **a.** Read questions 21 to 24 to develop a sense of what to focus upon when you read the article.
- **b.** Carefully read the article with these questions in mind. Remember to be an active reader by using a highlighter pen and/or by taking point-form notes. You can learn more about effective reading strategies by turning to "Reading for Understanding" in the Reference section.
- **c.** Save this evidence of your active reading strategies to help you answer questions 21 to 24.

21. The researchers used three tests to predict whether one of the men being studied was at risk of sudden death from a heart attack.

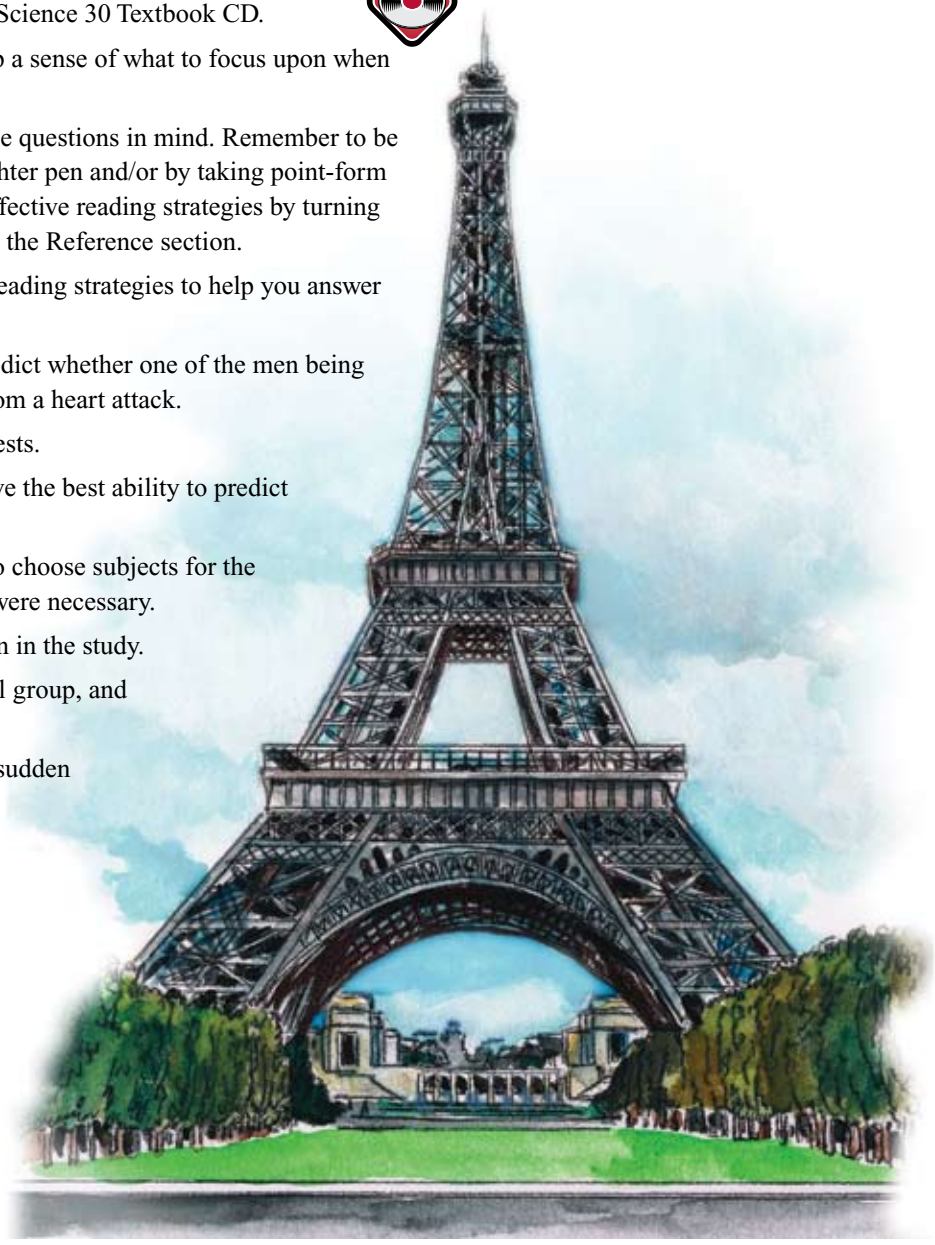
- **a.** Identify and describe each of the tests.
- **b.** Identify the test that appears to have the best ability to predict sudden death from a heart attack.

22. Describe screening procedures used to choose subjects for the study. Explain why these procedures were necessary.

23. The data was collected from 5713 men in the study.

- **a.** How many men were in the control group, and how was this group defined?
- **b.** How many men in the group died sudden deaths from heart attacks?

24. List some limitations of this research.



Chapter 2 Genetics

It's a beautiful fall day, and the young family in the photograph is getting ready to join their extended families for a big barbecue. For the girl, Roxanne, the barbecue will top off a near-perfect day at her grandparent's place where she got to play with her cousins and explore the farm. All day long Roxanne heard from different relatives about how much she is like her mother or how similar she is to her father. In terms of her physical traits, if you look closely at the photograph, you can see that Roxanne inherited some features from her mother and some from her father. The colour of Roxanne's hair and skin seem to be inherited from her father, while the colour and shape of her eyes are attributable to her mother.

How is it determined what traits are inherited from each parent? Is it true that some traits from one parent are "overpowered" by traits from the other parent? Does chance play a role in determining which traits are passed on?

Physical traits and many other characteristics are passed on from parents to offspring as information encoded in long molecules within cells. Genetics is the study of information and instructions inside cells. A greater understanding of how these genetic instructions are written and inherited by offspring has been applied to help solve crimes, understand diseases, make new medicines, and even re-design living organisms.

By the end of Chapter 2 you will have studied some history and major principles behind the science of genetics. You will not only have looked at how some genetic diseases can arise from inherited traits, but you will have examined the effect of genetic mutations. Throughout the chapter you will look at some ethical considerations of genetics by assessing the risks and benefits of using several genetic technologies.

Try This Activity

Tongue Rolling

A combination of different physical characteristics makes each person unique. Many physical differences—such as the colour of eyes, hair, and skin—are due to slight differences in the genetic information that each individual received from his or her biological parents. One characteristic determined by your genetic information is the ability to roll your tongue. Your teacher will ask you and class members to each try to roll your tongues like the person in the photograph.

Discussion

1. Were you able to roll your tongue?
2. Are more people in your class tongue rollers or non-tongue rollers? Which is the most common characteristic?
3. Do you think that if a non-tongue roller practised a lot, this person could roll his or her tongue?
4. Why do you think some people are able to roll their tongues and others are not? Is there a benefit to being able to roll your tongue?
5. Speculate about the answers to the following questions.
   a. Can a child have the ability to roll her tongue even if neither biological parent is able to?
   b. Could two tongue-rolling parents have a non-tongue rolling child?

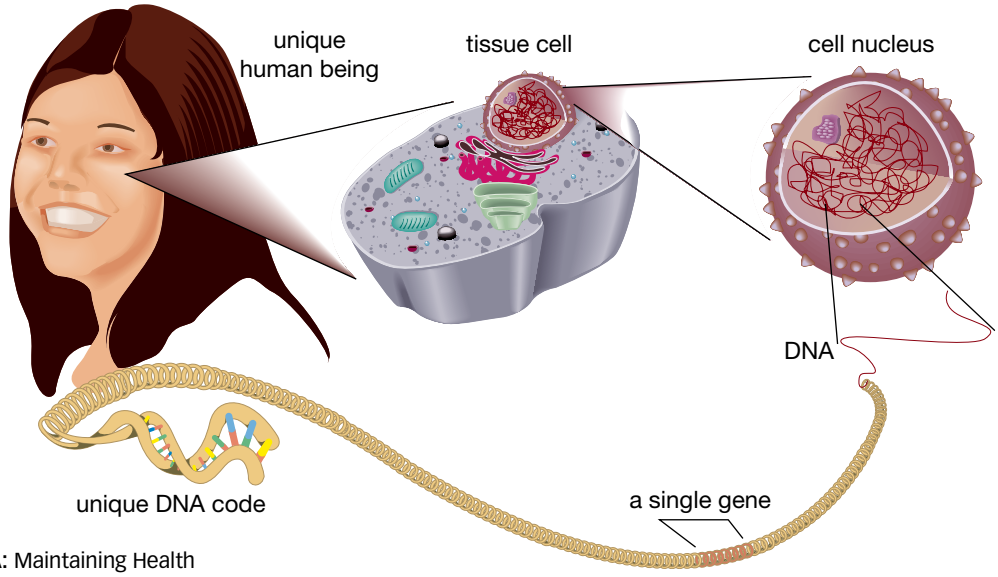
You will have an opportunity to move from speculation to writing well-explained answers when you return to some of these questions later in Chapter 2.

2.1 What Is Genetics?

People come in a wonderful variety of shapes, sizes, and colours. Some aspects of a person's appearance—such as hair style—are changeable and a result of personal grooming choices or current fashion trends. Traits that are not changeable include features such as the natural colour of your skin, your blood type, and whether or not you have dimples in your cheeks. Although it's easy enough to simply say that these traits are inherited, it is considerably more challenging to explain how a person's cells develop in a way that favours one trait over another.

If you were to use a microscope to look closely at any living thing, you would see that the organism is made up of small living units called cells. Some organisms are so tiny that they are made up of just one cell, whereas large and complex organisms are made up of trillions of cells. The cells in your body do all the jobs needed for you to live and remain healthy.

Locating Genetic Information Within the Cell



Chromosomes

In previous science courses you have studied the major parts of a cell. Recall that cells have a region called the nucleus that acts like a command centre to direct cell activity. If you used a very powerful microscope to look deep inside the nucleus of any one of your body's cells, you would find long strands of information called **chromosomes**. Each chromosome can be thought of as a book of instructions: almost like a cookbook full of recipes. A very simple organism, such as a bacterium, has all of its information on a single long strand. More complex organisms, such as people, tend to have several chromosomes. The number of chromosomes in a cell's nucleus depends on the type of species. Humans have 23 pairs of chromosomes or 46 total chromosomes. All the chromosomes in your nuclei are like a library of cookbooks or a complete set of instruction manuals with all the necessary information to run the activities of your cells.

chromosome: a strand of DNA that contains the instructions for making proteins

Chromosomes become X-shaped before cells divide.

Chromosomes are found in pairs. One outcome of this design is that a backup copy of important information is created. However, since one chromosome comes from each parent, there might be slight differences in the instruction subsets. To return to the cookbook analogy, it is like a cookbook with two recipes for chocolate fudge. Despite the fact that the instructions are nearly identical, slight variations in cooking times and temperatures result in food with different textures and consistencies.

Every cell has a complete set of chromosomes even though that particular cell might not need all the information to make a complete organism. The cell only uses the instructions required for its particular needs. This is just like several people who own the same cookbook using different recipes from the same book depending on their individual needs. If you made a cake from a cookbook recipe, you wouldn't have to read through the whole book each time you wanted to make a cake. In the same way, a particular cell from the pancreas only needs to read some of the information. This data could be, for example, a description of how to produce insulin.

Cookbooks and chromosomes differ in an important way. Information in cookbooks is organized by topic with all the recipes for desserts in one chapter or in one specific cookbook in your cookbook library, but the chromosome genes are not organized in a similar way. The instructions for making a hand are not found on one particular chromosome, but the instructions are instead spread among several chromosomes.

Chromosomes are generally long, thin strands that are coiled at regular intervals around protein molecules for protection. Chromosomes are best seen and photographed when the cell is dividing. At this time, each chromosome produces an identical copy of itself. The two copies, which remain attached at one point, shorten by coiling to produce the characteristic X-shape shown in photographs.

Genes

At specific places on each chromosome there are encoded instructions called **genes**. If a chromosome is like a recipe book, then a gene is like a specific recipe that provides the detailed instructions for building certain proteins. In order for instructions to be stored, communicated, and then used to complete a task, a language is necessary. Cookbooks are written in languages—like Korean or German—that people can understand. Genetic instructions are written in a chemical language called **deoxyribonucleic acid** or **DNA** for short. This language is encoded in a molecule. DNA has a distinctive shape, called a double helix, that looks like a twisted ladder or a spiral staircase. The sequence of chemical components in the DNA molecule encodes information. Each rung in the spiral ladder of the DNA molecule can be thought of as an individual letter in the cookbook.

- **gene:** a segment of DNA that carries instructions that result in the production of proteins
- **deoxyribonucleic acid (DNA):** the twisted ladder-shaped molecule that contains the genetic information of cells
- **genetics:** the science of gene function and inheritance

The ability to roll your tongue is controlled by a gene and determined by a certain pattern or sequence along a DNA molecule. People who are unable to roll their tongues do not have the gene instructions that allow the tongue muscles to roll. Even though two people each have the gene instructions for making hair or an eyeball, slightly different recipes create curly hair instead of straight hair or blue eyes rather than brown eyes. The combination of instructions from the genes on your chromosomes determines many of your characteristics. **Genetics** is the scientific study of how genes work to determine characteristics and to resolve how genetic information gets passed from parent to offspring.

Practice

1. **a.** Describe two ways in which chromosomes and cookbooks are similar.
   b. Describe one way that chromosomes and cookbooks are different.
2. How many chromosomes are found in one of the cells in your hand?
3. Why do chromosomes often appear with an X-shape in diagrams and in photos taken through a microscope?
4. Describe what is meant by the term *double helix*.

Human Karyotype

- **karyotype:** an image that organizes the chromosomes of a cell in relation to number, shape, and size
- **centromere:** the region on a replicated chromosome that attaches the two identical copies during cell division

The human body contains 23 pairs of chromosomes, but the chromosomes are not neatly organized in the nucleus. In fact, they are in a jumble that looks like a plate of spaghetti. A **karyotype** is an image of all the chromosomes in one nucleus that have been matched up into their respective pairs and arranged from the largest pair to the smallest pair. A karyotype allows geneticists to better study the chromosomes in a nucleus. Geneticists use three features to identify and match up chromosomes:

- the length of the chromosome (The longest chromosome is numbered as chromosome 1, etc.)
- the pattern of dark bands produced on each chromosome when they are stained
- the position of the chromosome's constricted part, called the **centromere**, which plays a role during cell division

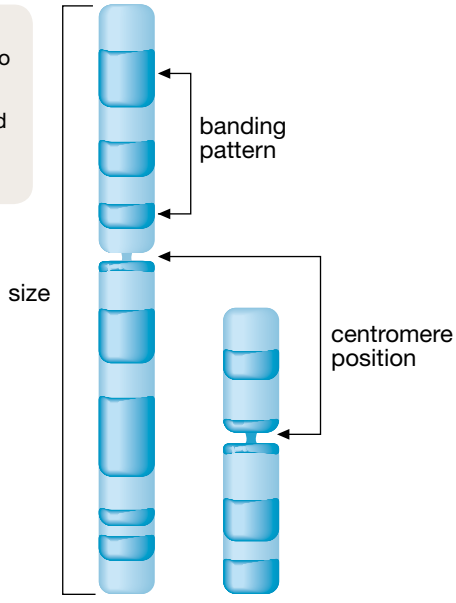


Figure A2.1 illustrates how it is possible for chromosomes to be matched up.

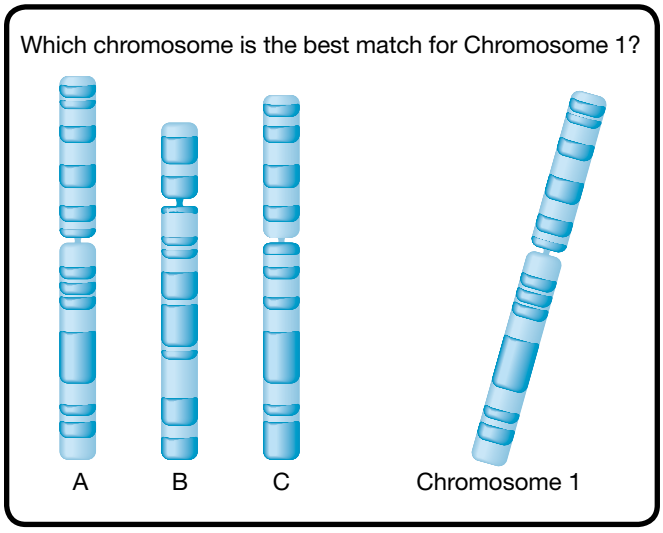
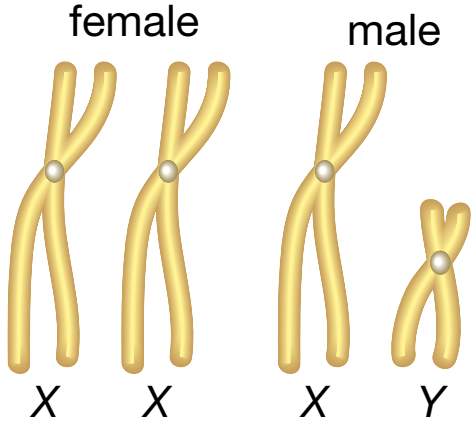


Figure A2.1

An artist's representation of the chromosomes makes the process of chromosome matching look easy—A is clearly the best match for Chromosome 1. However, there are complications when examining photographs of actual chromosomes taken through a microscope—it is challenging to interpret the somewhat grainy photographs of these incredibly tiny objects. Also, the last two chromosomes in the karyotype are called the *sex chromosomes* because they determine the organism's gender.

The pair of chromosomes matched at chromosome 23 determine the sex of the individual.



Note that these chromosomes are shown after replication.

If an individual has two *X* chromosomes, she is a female. If an individual has one *X* chromosome and one *Y* chromosome, he is a male. Since the *Y* chromosome is considerably smaller than the *X* chromosome, this creates an extra challenge for matching chromosomes for a male.

As you'll discover in the next activity, overcoming these challenges is like sorting out a jigsaw puzzle.

Try This Activity

Make a Human Karyotype

To complete this activity you will need a copy of the handouts "Cut and Paste Karyotype Activity" and "Cut and Paste Karyotype Activity—Matched," which are available on the Science 30 Textbook CD.

Science Skills

- ✓ Performing and Recording
- ✓ Analyzing and Interpreting

Purpose

On the handouts you will find the corresponding chromosome that best matches each of the numbered chromosomes to form a human karyotype.

Materials

- "Cut and Paste Karyotype Activity" handout
- "Cut and Paste Karyotype Activity—Matched" handout
- scissors
- glue or transparent adhesive tape

Procedure

step 1: Cut out the 23 unnumbered chromosomes from the handout titled "Cut and Paste Karyotype Activity."

step 2: Place each unnumbered chromosome with a numbered chromosome to produce a matched pair.

step 3: Check your completed karyotype by using the handout named "Cut and Paste Karyotype Activity—Matched."

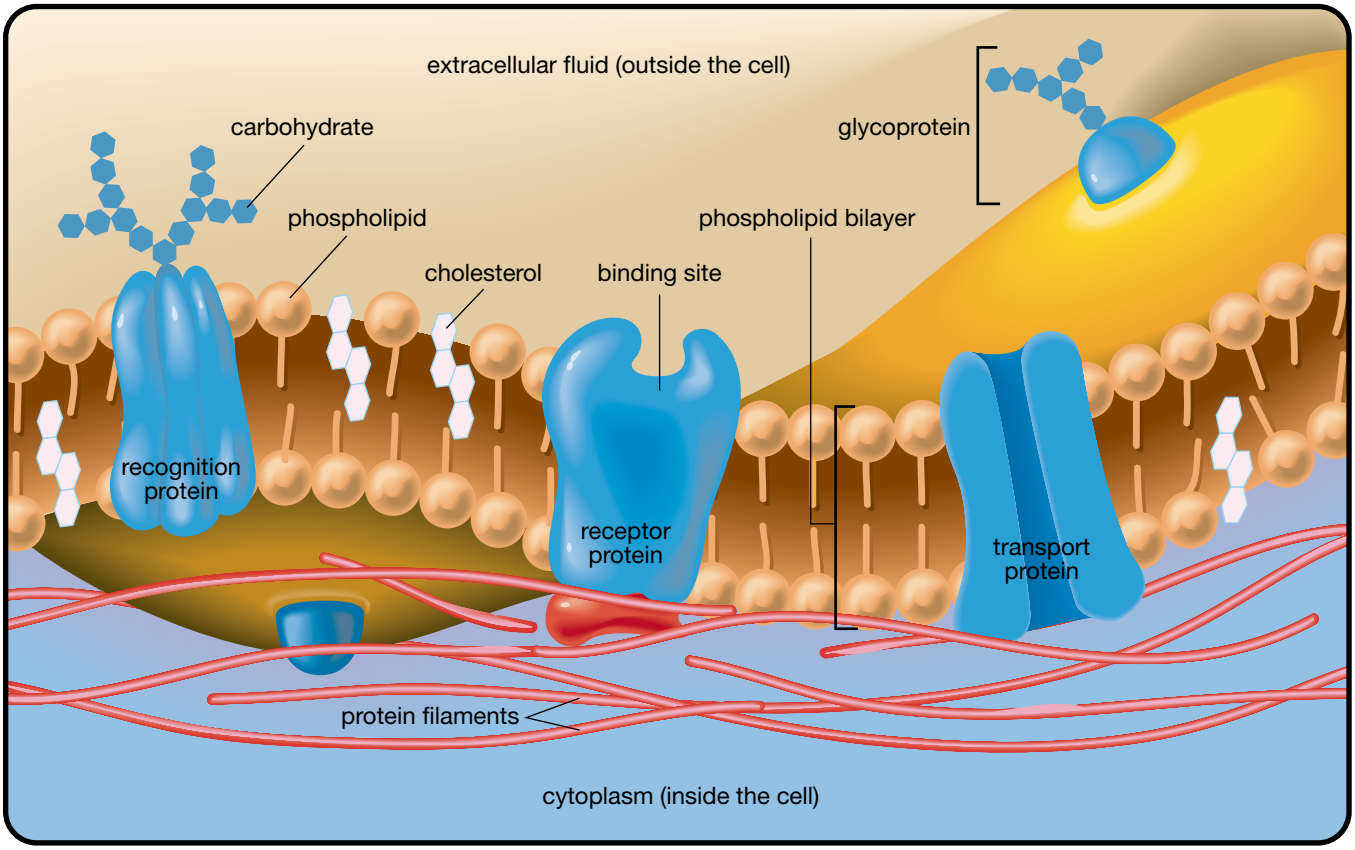
Analysis

1. Why it is beneficial to have two sets of chromosomes? Where does each set of chromosomes come from?
2. **a.** Describe what is different in terms of shape and size in the last pair of chromosomes.
   b. Is this individual a male or a female?
3. Explain how a karyotype of an individual might be useful to scientists.

The Role of Proteins

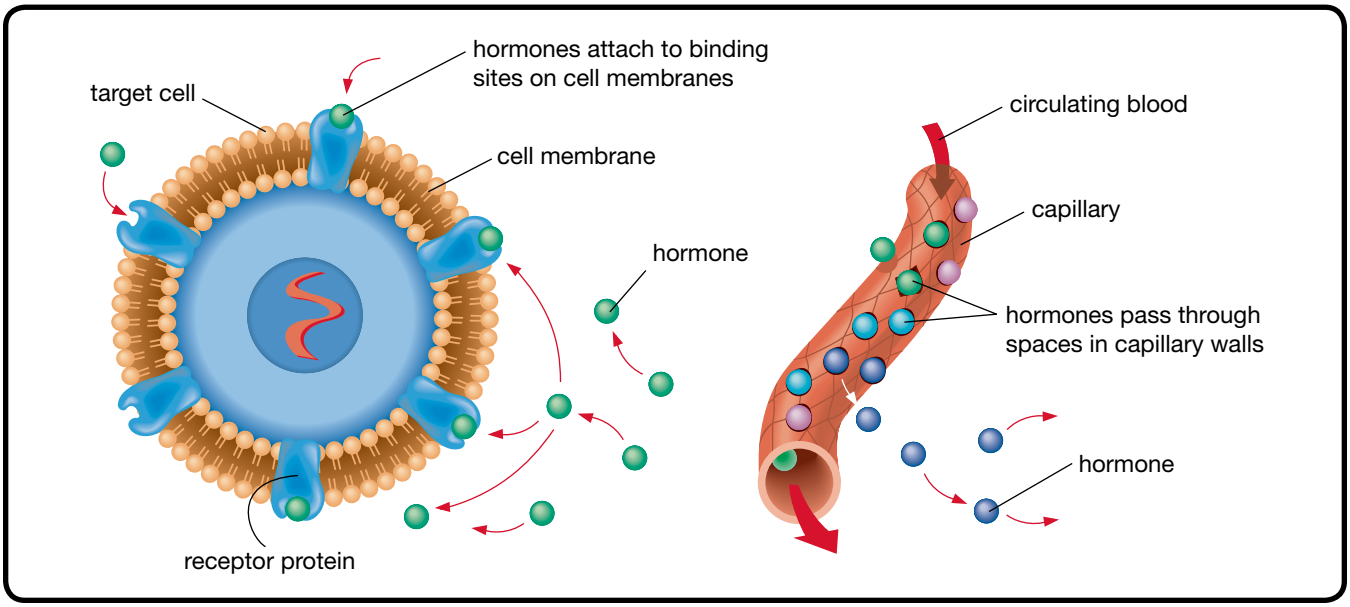
More than 25 000 genes are spread over the 46 chromosomes in the human karyotype. Chromosomes are like cookbooks, and each gene is a recipe for making a specific protein. This means there are more than 25 000 different recipes for proteins. Why is all this vast library of genetic information focused on making proteins? What role do proteins play in the body?

Cell Membrane Proteins



Proteins are molecules that have versatile and important bodily functions. If you took away all the water in the human body, about 50% of the dry mass left behind would be proteins. The human body produces tens of thousands of proteins, each with a unique structure and a specific job. If the body is like an engine, each protein is like a tiny specialized part that completes a specific task to keep the engine running. Hormones are proteins that co-ordinate and regulate the body's activities. This is shown in Figure A2.2.

Figure A2.2



The major types of proteins found in organisms and some of their roles are summarized in the following table.

SUMMARIZING THE ROLES OF PROTEINS

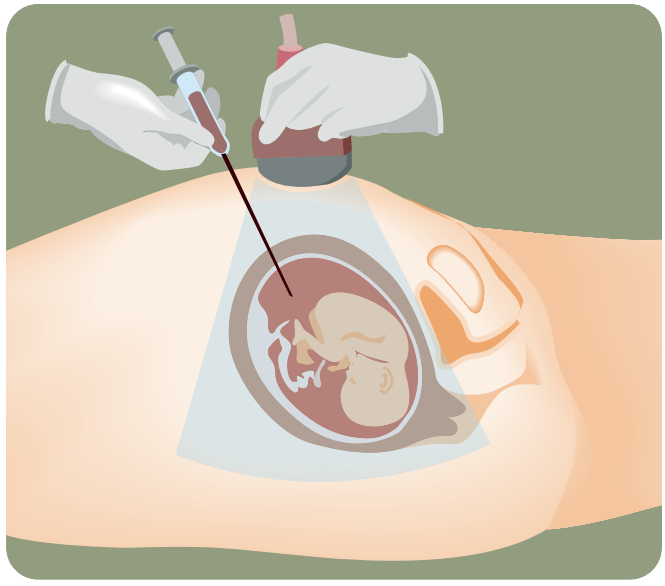
Type of Protein	Role of Protein	Example
enzyme	Enzymes speed up chemical reactions where molecules are broken apart or put together.	Amylase is a digestive enzyme in your saliva that breaks down long starch molecules into shorter, more digestible glucose molecules.
structural	Structural support and frameworks are created to attach to other proteins.	Keratin is a structural protein that makes up your hair and nails. Collagen is a structural protein that provides a framework for skin and internal organs.
transport	Materials are moved within the cell or body.	Cell membrane proteins form channels and pumps in the cell membrane to help needed materials flow into the cell and unwanted materials flow out of the cell.
hormone	Hormones act as signals to co-ordinate and regulate activities in the body.	Insulin is a hormonal protein that regulates blood sugar. Insulin is produced in the pancreas and moves in the bloodstream to other organs to influence their use of glucose.
contractile	Contractile proteins change shape and can create larger movements when they work together.	Actin and myosin are proteins that band together to allow muscles to contract.
defensive	Defensive proteins protect the body against disease.	Antibodies are proteins that act in the body by attaching to disease-causing pathogens and foreign material.
energy	Energy proteins serve as a source of chemical potential energy that can be released by its decomposition.	Casein is an energy protein found in milk.

Practice

5. Identify examples of the kinds of molecules genes are designed to produce.
6. Describe the function of proteins in cell membranes.

Amniocentesis

An **amniocentesis** is a prenatal test done to look at the karyotype of an unborn child. During the test, a small amount of amniotic fluid is drawn out with a large needle from the area around the fetus. An image produced with ultrasound helps to direct the path of the needle. Some of the developing baby's cells are floating in the amniotic fluid and these cells are examined for genetic abnormalities, such as the presence of additional chromosomes, as is the case in Down syndrome. Amniocentesis is not routinely offered to expectant mothers because the procedure can slightly increase the chance of miscarriage, and it is only performed in pregnancies where there is a high risk of genetic diseases or deformities. Factors that can increase the risk of genetic diseases or deformities include the mother's age (above 40) or a history of severe diseases known to have a genetic origin.



amniocentesis: a prenatal test done to look at the karyotype of an unborn child

Making More Chromosomes

When a cell divides, it must provide genetic information to each of the new cells that form from the cell division. This means that exact copies must be made of the long strands of DNA within each of the chromosomes. Depending upon the type of cell, there are two basic ways in which this process can occur. These methods are mitosis and meiosis.

Mitosis

In order to grow and to replace cells that are dead or damaged, your body must constantly make new cells. Skin cells, for example, need to be frequently replaced by new cells. A body cell, or an **autosomal cell**, divides by growing large and making extra copies of all its parts and then splitting into two. An autosomal cell is shown at the top of Figure A2.3. Instead of showing 23 pairs of chromosomes, the simplified illustration shows only one pair. Note that one chromosome from the pair is inherited from the father and the other from the mother. Since each chromosome within the pair carries genes for the same characteristics at the same chromosome location, the pair of chromosomes are called **homologous chromosomes**.

To ensure that new cells have the necessary genetic information, autosomal cells must make a copy of their chromosomes before dividing. It is said that the DNA **replicates**. The replicated chromosomes attach at the centromere to form a distinctive X-shape. If each of these chromosomes is thought of as a recipe book, the process of DNA replication increases the number of copies of each book from two to four.

After being replicated, the chromosomes move to line up along the cell's middle or equator. The duplicate strands of the chromosomes are then pulled apart. The cell membrane pinches in to split the cell into two new cells with two sets of chromosomes called **daughter cells**. The process of cell division in autosomal cells is called **mitosis**. Note that with two copies of each chromosome, daughter cells are identical to the original autosomal cell that began the whole process. Biologists refer to the original cell and the daughter cells as **diploid cells** because each of these cells has two copies of each chromosome type.

Some organisms are able to produce a new organism by mitosis. This is called **asexual reproduction**. Offspring produced asexually are genetically identical to their parents. Bacteria reproduce asexually by simply splitting in two. A strawberry plant or a spider plant often makes a small copy of itself that breaks off or shoots off on a runner and grows into a new plant. Growing this small copy is called *budding*. The ability of plants to reproduce asexually allows people to use plant cuttings to grow into a whole plant. This plant is a genetic copy of its parent plant.

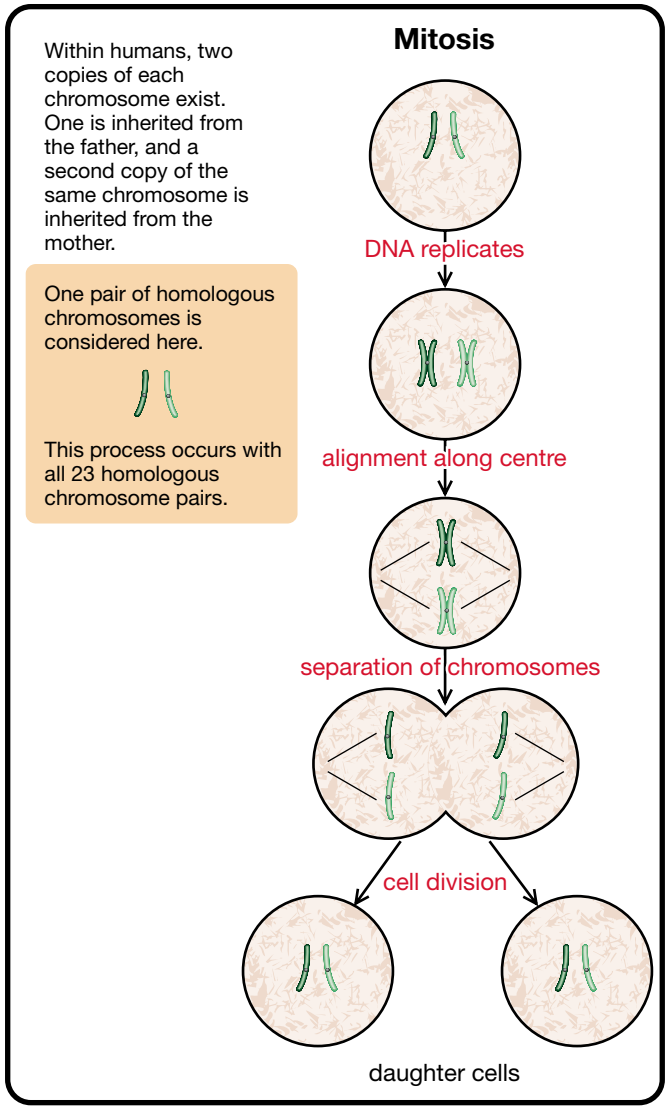


Figure A2.3

- **autosomal cell:** a cell of the body not involved in sexual reproduction
- **homologous chromosomes:** a pair of chromosomes that would be matched during karyotyping because they have the same length, centromere position, and staining pattern
- **replicate:** to produce an exact copy of a DNA strand
- **daughter cells:** the two identical cells produced during mitosis
- **mitosis:** the division of an autosomal cell into two identical daughter cells
- **diploid cells:** cells with pairs of homologous chromosomes
- **asexual reproduction:** the production of genetically identical offspring from one individual

Meiosis

Most organisms do not reproduce asexually. Instead, they produce special reproductive cells called sex cells or **gametes**. These cells combine to make new and unique offspring. In animals the gametes are called sperm and egg, and in plants they are called pollen and egg. The process of producing gametes, called **meiosis**, begins in the same way as mitosis. Cells in the reproductive organs start with chromosomes in homologous pairs. Again, instead of showing 23 pairs, only one pair is noted in Figure A2.4. The DNA replicates and the replicated chromosomes form the characteristic X-shape.

Meiosis begins to differ from mitosis in the steps that follow. The first difference that occurs is that homologous chromosomes pair up and exchange parts. Some DNA segments of the chromosome from one parent are exchanged for corresponding DNA segments on the other parent's chromosome. This process of exchanging genetic material during meiosis is called **crossing over**. Since the pattern of DNA has been altered, crossing over creates slight genetic differences in the chromosomes.

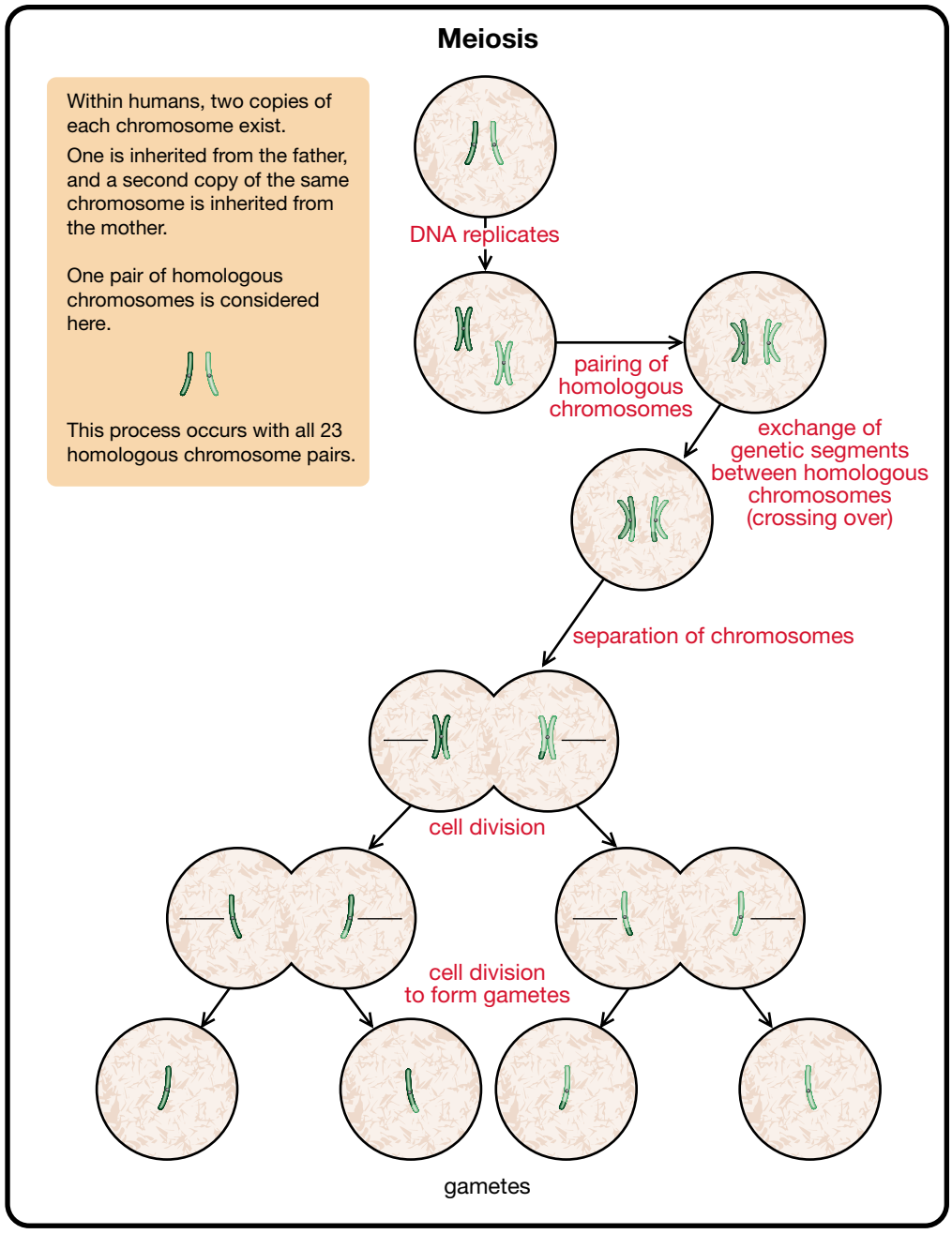


Figure A2.4

After crossing over, chromosomes align along the centre of the cell so that the homologous chromosomes are side by side. The homologous chromosomes separate and two daughter cells are produced. Next, a second cell division occurs, resulting in four cells that are called gametes.

Compare the number of chromosomes in the gametes with the number in the original cell at the top of Figure A2.4. Since there was no DNA replication prior to this second cell division, the four gametes each carry only one set of chromosomes rather than two sets in autosomal cells. The gametes are referred to as **haploid cells**, as they only have half the number of chromosomes as do autosomal cells.

- **gamete:** a sex cell, such as a sperm and an egg, produced during meiosis with only one copy of each chromosome type
- **meiosis:** a two-stage form of cell division that produces gametes with only half of the number of chromosomes as the original cell
- **crossing over:** the exchange of corresponding segments of DNA between maternal and paternal chromosomes during meiosis
- **haploid cell:** a cell that has only one member from each pair of homologous chromosomes

In people there are 23 different types of chromosomes. Biologists use the shorthand $n = 23$ to communicate this idea. Each gamete, either a sperm cell or an egg cell, has only one copy of each of these chromosomes. The notation for these cells is 1n, meaning one copy of each type of chromosome.

Each of the other body cells—the autosomal cells—has two copies of each type of chromosome. These cells are described as 2n cells, meaning two copies of each chromosome. The following memory device can help you remember these ideas.

Mitosis is remembered as "mi two sis."	**Meiosis is remembered as "mei one sis."**
• produces diploid cells (**2n**), with **t**wo copies of each chromosome	• produces haploid cells (**1n**), with **o**ne copy of each chromosome

Practice

7. Obtain a copy of the handout "Mitosis" from the Science 30 Textbook CD.

- **a.** Add the missing labels to "Mitosis."
- **b.** Add the labels "1n" and "2n" to describe the original cell and the daughter cells.

8. Obtain a copy of the handout called "Meiosis" from the Science 30 Textbook CD.

- **a.** Add the missing labels to this diagram.
- **b.** Add the labels "1n" and "2n" to describe the original cell and the gametes.

Utilizing Technology

Comparing Mitosis and Meiosis

Background Information

Now that you have seen the details of mitosis and meiosis, it's important to be able to keep the big picture in mind and not to lose sight of the main ideas. This activity provides an opportunity to reinforce essential concepts about mitosis and meiosis.

Science Skills

✓ Performing and Recording

Purpose

You will use an applet titled "What Is Mitosis/Meiosis?" to reinforce the essential concepts that are necessary to understand mitosis and meiosis.

Procedure and Observations

step 1: Locate the applet "What Is Mitosis/Meiosis?" from the Science 30 Textbook CD.

step 2: Review the list of analysis questions. Then watch the applet.

step 3: Watch the applet again, only this time use the natural pauses between screens to record your answers.

Analysis

1. A human being begins as one cell and then grows into a body of a hundred trillion cells.

- **a.** Define the term *diploid cell*.
- **b.** Describe the arrangement of chromosomes in human body cells.
- **c.** Explain why it is necessary, before it divides, for a body cell to make a copy of each chromosome.
- **d.** Identify the process that describes how one cell becomes a body of trillions of cells.

2. Not all cells in the human body are diploid cells.

- **a.** Define the term *haploid cell*.
- **b.** Describe in general terms the process that produces haploid cells.
- **c.** Identify the name of the process that you described in question 2.b.
- **d.** Explain the purpose of haploid cells.
- **e.** Explain what happens when two haploid cells combine.

3. Describe the essential difference between mitosis and meiosis.

Fertilization

When chromosomes in a male gamete join up with chromosomes in a female gamete during fertilization, the fused cell contains the two sets of chromosomes found in an autosomal cell. With two sets of chromosomes, the fertilized egg is able to grow and develop through the process of mitosis.

The advantage of sexual reproduction is that diverse offspring are produced. The genetic combination of chromosomes from each parent results in offspring with the possibility of different traits. As shown in Figure A2.5, even with a simplified model using only one pair of chromosomes, there are 16 possible outcomes. If the full complement of 23 pairs of chromosomes are used, there will be more than 70 trillion possible ways for gametes from a mother and a father to join and create a new human being. The number of possibilities would be even greater if crossing over was considered. Given that this number is more than all the people who have ever lived on planet Earth, unless you are an identical twin, there has never been anyone with your exact DNA. In other words, you are a unique creation.

Fertilization

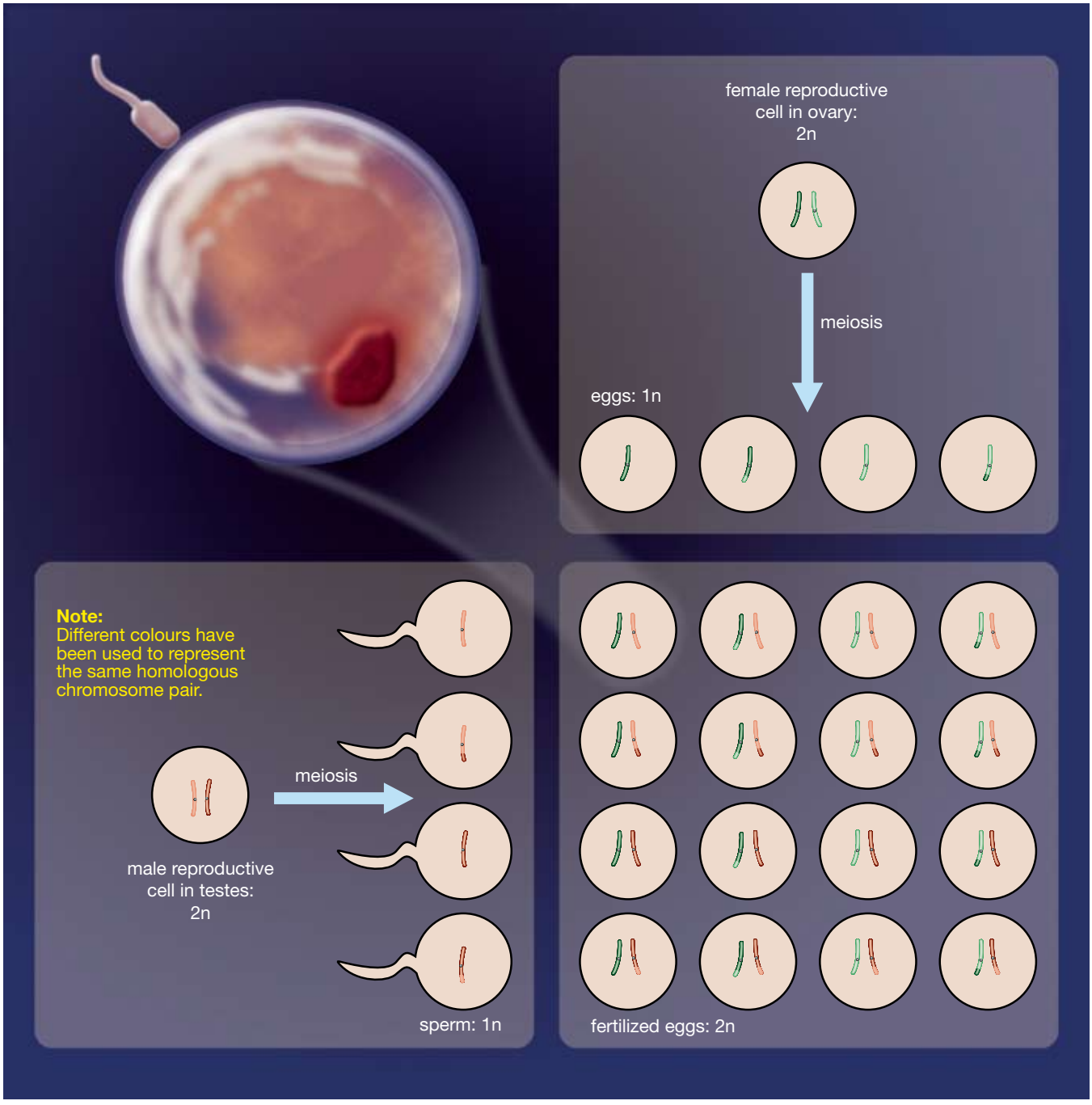


Figure A2.5: This simplified model shows the outcomes from 1 pair of chromosomes instead of 23 pairs.

Sexual reproduction is clearly a valuable mechanism for increasing the variety among organisms in a population. Note the supporting role that crossing over plays to increase variety in the gametes produced by meiosis. The exchange of genetic material during crossing over further increases the genetic diversity of the offspring, and this is part of the reason why two siblings from the same parents can look quite different. Genetic diversity is important because a genetically diverse population is less susceptible to disease.

Utilizing Technology

Determining Numbers of Unique Offspring

Purpose

You will use a spreadsheet to verify the following statement:

> " . . . there would be more than 70 trillion possible ways that gametes from a mother and a father can join to create a new human being."

Science Skills

✓ Analyzing and Interpreting

Procedure

You may complete this activity by using a spreadsheet or a graphing calculator. Use the following template as a guide. Continue the patterns shown until the Number of Pairs of Chromosomes column reaches 23 pairs. At this point, the table is complete.

Number of Pairs of Chromosomes	Number of Possible Gametes Produced by One Parent (excluding crossing over)	Number of Possible Unique Combinations of Offspring Produced by Two Parents
n	2^n	$(2^n) \times (2^n)$
1	$2^1 = 2$	$(2^1) \times (2^1) = 2 \times 2 = 4$
2	$2^2 = 4$	$(2^2) \times (2^2) = 4 \times 4 = 16$
3	$2^3 = 8$	$(2^3) \times (2^3) = 8 \times 8 = 64$
4	$2^4 = 16$	
5		

Analysis

If the number of gametes produced by each parent is 2^n, explain why the number of offspring produced by two parents is $(2^n) \times (2^n)$.

Selective Breeding

Long before chromosomes and genes were discovered, people recognized that offspring had a similar appearance to their parents and hypothesized that characteristics could be inherited. However, people were not able to explain how these characteristics were passed on from parents to offspring. They lacked necessary tools—such as microscopes powerful enough to see cells and their parts—to understand mechanisms for the inheritance of traits or the genes responsible for them.

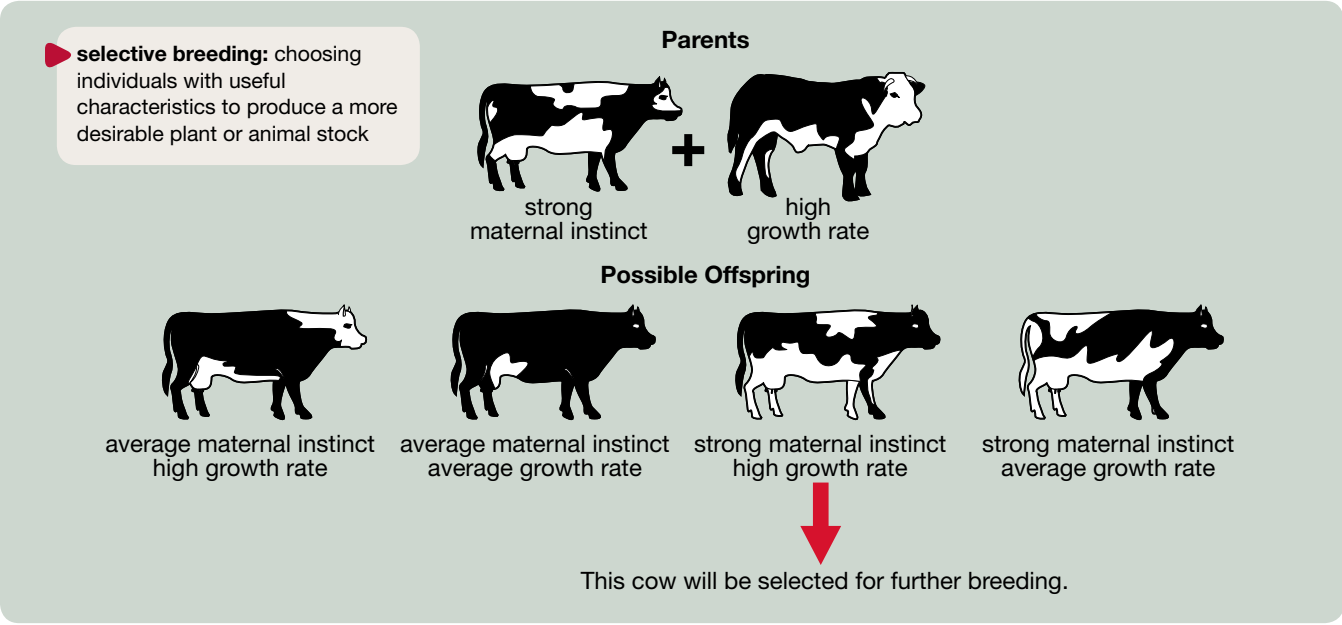


Figure A2.6: Raising cattle for beef production involves selective breeding. A strong maternal instinct in females means good nurturing and feeding of calves. Orphaned calves often grow poorly or they may die. A high growth rate demonstrates that the animal's metabolism is able to convert ingested food into tissue (meat).

Ancient peoples who lived in aristocratic societies believed that the monarchs passed on their noble or royal blood to their children. These people also realized that they could select individuals with desirable characteristics from their domestic crops and livestock. These crops and livestock could then be bred to produce offspring possessing desirable traits. Farmers could also prevent individuals with undesirable characteristics from breeding. The practice of **selective breeding** allowed farmers to create stocks that met their particular needs.

Figure A2.7: Corn husks were used by Iroquois to make masks or false faces that had special spiritual significance in healing ceremonies.

First Peoples of North America and Central America employed selective breeding to grow domesticated crops—such as maize (corn)—for thousands of years before Europeans arrived in the area. In 1612, French explorers around the Great Lakes recorded that Haudenosaunee (Iroquois) people made popcorn by putting maize kernels in pottery full of heated sand. It is believed that the maize First Nations people grew is a result of centuries of selective breeding of teosinite, a wild grass. The corn and popcorn that you eat today is descended from the breeds developed by First Nations people.

Practice

Use the following information to answer question 9.

In previous courses you studied the theory of natural selection. This theory states that evolution takes place because more organisms are produced than can survive and that only the organisms best suited to their environment survive to reproduce and, in turn, pass on their advantageous traits to their offspring. According to this theory, the environment determines what organisms will be successful and will therefore have their traits determine the characteristics seen in future generations.

9. a. Describe how selective breeding differs from the process of natural selection.

b. Describe how selective breeding is similar to the process of natural selection.

10. Genetic engineering can be defined as the manipulation of genes in organisms to produce desirable characteristics and to eliminate undesirable characteristics.

a. Explain how selective breeding is a form of genetic engineering.

b. Identify the first people to practise genetic engineering in Canada.

Early Ideas of Inheritance

The development of simple microscopes allowed for the discovery of cells, such as sperm and eggs; but these instruments were not powerful enough to observe chromosomes. During the seventeenth and eighteenth centuries, the theory of preformation was popular. Preformation is the idea that all body parts are already formed at the beginning of development and simply grow from a very tiny full-formed body to a larger body. Prominent scientists including Anton van Leeuwenhoek and Marcello Malpighi, the person who discovered capillaries, were supporters of the preformationist theory. However, there was some debate about whether the pre-formed body was found in the sperm or the egg.

Figure A2.8: Charles Darwin

In the nineteenth century, Charles Darwin used his studies of the natural world to develop and publish his theory of evolution. His theory stated that organisms best suited to their environment survived to pass on their characteristics to their offspring, but Darwin was not able to explain how characteristics are passed on.

Discovering Genes

In the 1860s, Gregor Mendel was the first person to undertake detailed and systematic studies into the inheritance of characteristics. Mendel was a monk who became interested in the breeding of pea plants that grew in the monastery where he lived. Little did he know that he would become known as "the father of genetics."

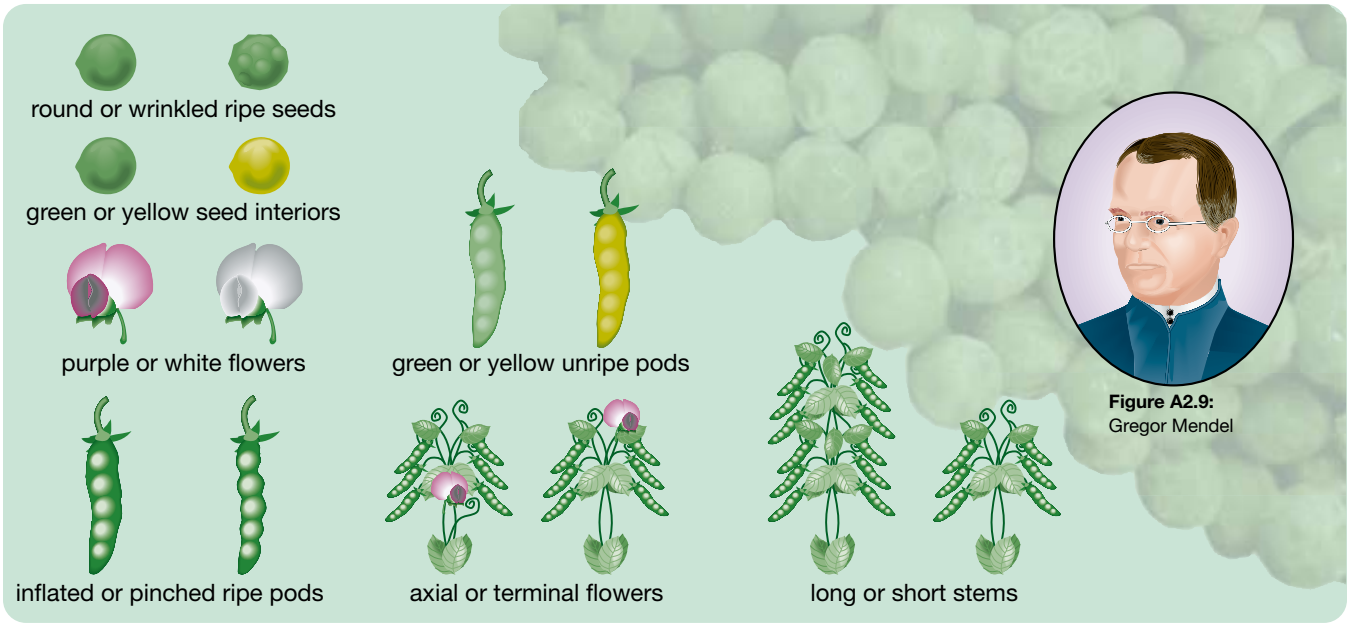


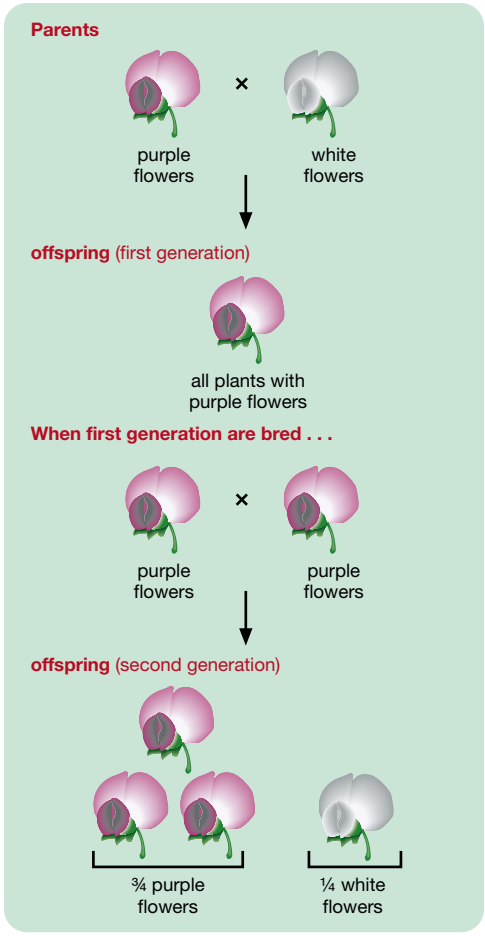
Figure A2.9: Gregor Mendel

Mendel noticed that certain inherited characteristics were not blended. For example, a pea plant with white flowers when bred with a pea plant with purple flowers did not produce offspring with blended characteristics. Instead, the offspring had either purple flowers or white flowers.

Common garden pea plants were good test subjects for his research because not only are they easy to grow in large numbers, but their reproduction can be manipulated by transferring pollen from one plant to another in a process called **cross-pollinating**. Mendel discovered that several traits in pea plants were easy to recognize and occurred in only one of two distinctive forms—purple flowers or white flowers, round seeds or wrinkled seeds, yellow seeds or green seeds, and other forms.

- **cross-pollinate:** transferring pollen between genetically different plants
- **self-pollinate:** transferring pollen from one plant to the female part of the same plant or to another plant with the same genetic makeup

Mendel observed that when he cross-pollinated a white flowering plant with a purple flowering plant, the offspring from this cross all had purple flowers. The white-flowered trait had disappeared in the generation of pea plants produced by this cross. When he bred individuals from this first generation, or allowed them to **self-pollinate**, he was surprised to see that the white-flowered trait returned in about a quarter of the second-generation's population. Experiments on other pea traits provided the same results. One trait completely disappeared in the first generation and then re-appeared in about a quarter of the second-generation plants.



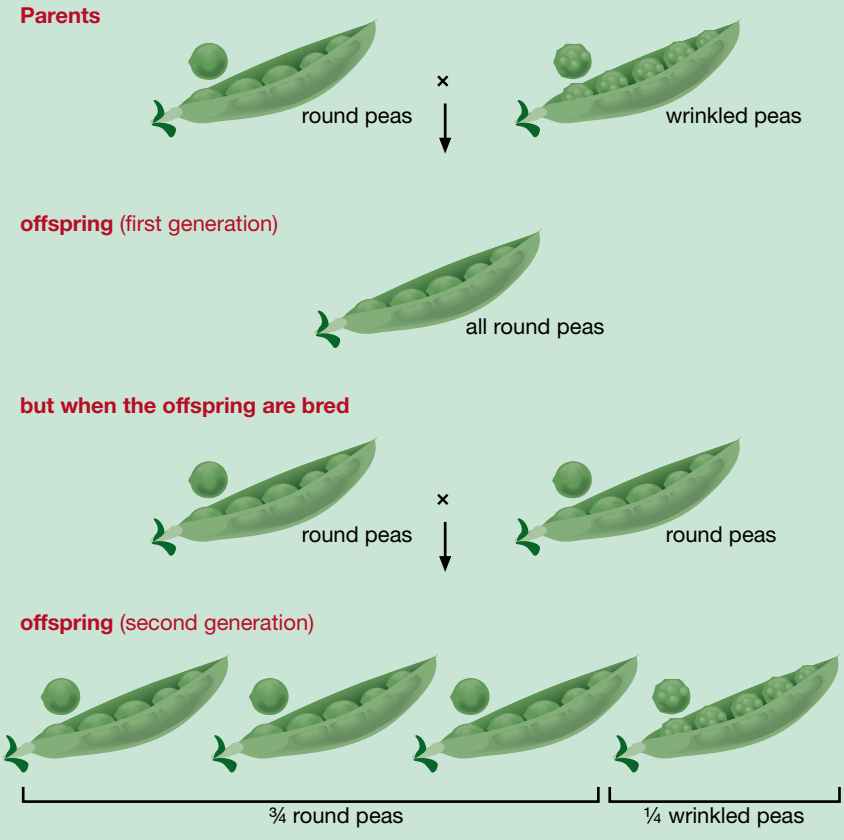
Mendel's Conclusions from His Pea Plant Experiments

(1) The inheritance of traits must be determined by factors (now called genes).

(2) Individuals randomly inherit one factor (copy of a gene) from each parent.

(3) Factors or genes are independently passed to the offspring. This means that a pea plant's inheritance of the gene that makes purple flowers does not affect the plant's inheritance of other traits, such as seed colour.

(4) Some genes are more powerful than others; so a trait may not appear in an individual because the more powerful gene masks it (e.g., purple flowers over white flowers), but it can still be passed onto the individual's offspring.



Even though Mendel was studying pea plants, he actually described the basic principles of genetic inheritance for all complex life forms, including people. Mendel published his results in 1866. Sadly, in his lifetime Mendel's results were not understood and the importance of his work was unrecognized. Sixteen years after Mendel's death, chromosomes were discovered and it was suggested that Mendel's factors might be carried on the chromosomes. After his death, Mendel's experiments were repeated by other scientists and his ideas and important contributions to the understanding of how traits are passed from parents to offspring were acknowledged.

2.1 Summary

A chromosome is a long strand of information made up of a double helix molecule called deoxyribonucleic acid or DNA. Chromosomes are found in pairs in the nuclei of autosomal cells. Genes are specific regions along the chromosomes and have the instructions for making proteins. Genetics is the study of how genes work to determine characteristics and how they get passed on.

A karyotype is an image of all the chromosomes paired up and organized for study by geneticists. The human karyotype is composed of 46 chromosomes, 22 pairs of autosomes, and 1 pair of sex chromosomes. The sex chromosomes determine the gender of an individual. Two *X* chromosomes produce a female, and one *X* chromosome combined with one *Y* chromosome produces a male.

Body cells, or autosomal cells, divide for an organism to grow or replace cells through the process of mitosis. During mitosis, two identical daughter cells are produced with identical sets of chromosomes. Some organisms can reproduce asexually through mitosis alone.

Sex cells, or gametes, are produced for sexual reproduction through the process of meiosis. During meiosis, four gametes are produced—each has half the number of chromosomes of an autosomal cell. The combination of male and female gametes creates a fertilized egg with the required two complete sets of chromosomes.

Selective breeding is a technology that has allowed people to develop more useful breeds of domesticated plants and animals to suit human needs.

Mendel's study of pea plants led to the development of a theory proposed where traits were inherited by factors (later called genes) and in which offspring randomly received a copy of a gene for each trait from its parents. Mendel also stated that possessing a gene for one trait did not affect the genes for other traits and that some genes were able to mask the other forms of an inherited trait.

2.1 Questions

Knowledge

1. Match the following terms with the analogy that best describes each term.

- DNA
- gene
- protein
- chromosome
- karyotype

a. a cookbook with several recipes in it

b. an entire library of cookbooks neatly arranged in order from the largest book to the smallest book

c. the cake produced by following recipe instructions

d. the letters and words in a recipe

e. the instructions for making a cake

2. How many chromosomes are in a normal human autosomal cell? How many chromosomes are in a normal human gamete?

3. Determine the gender of an individual who has two *X* chromosomes in each autosomal cell instead of an *X* and a *Y* chromosome.

4. Dogs have 78 chromosomes, cats have 34 chromosomes, and goldfish have 94 chromosomes. Explain why the usual number of chromosomes in autosomal cells for any species is always an even number.

5. Describe what would happen if the process of meiosis did not occur and two cells with two sets of chromosomes combined to produce a new child. Why is meiosis necessary?

6. Identify the term Mendel used instead of *gene*.

7. List some reasons why pea plants are so well suited for genetic studies.

Applying Concepts

8. Obtain a copy of the handout "Comparing Mitosis and Meiosis," from the Science 30 Textbook CD.

a. Without looking at information from this lesson, attempt to add the missing labels to this diagram.

b. Biologists use the letter *n* to represent the number of different chromosomes in a cell. Label the cells at the top and bottom of the handout either 1n or 2n.

c. Use the information in Lesson 2.1 to check and correct your answers to 8.a.

9. Sexual reproduction creates beneficial genetic diversity. List some possible advantages of asexual reproduction.

10. Explain the steps you would take in the process of selective breeding to create a fast-running breed of dog.

11. Propose some reasons why van Leeuwenhoek and other scientists of his time believed in the preformation theory.

2.2 Inheritance

- **acquired traits:** traits acquired during a person's lifetime because of experiences, education, and upbringing, such as a scar from a cut or the ability to speak a particular language
- **inherited traits:** traits genetically passed on from one generation to the next, such as a particular blood type or eye colour

You have probably been told that you have characteristics similar to another member of your family. Perhaps someone has said that you have your mother's hair, your father's eyes, or that you inherited a trait from one of your grandparents. You might have compared your own characteristics to other family members or wondered which traits you might someday pass on to your children.

Although people have long understood that characteristics are inherited from their parents, they did not understand the mechanisms that enable inheritance to happen. As Mendel's work became more well known and understood, scientists were able to use his observations, terminology, and the results of his experiments to make predictions about how and what characteristics are passed on to offspring.

The discovery of genes and the field of genetic research has helped to answer questions concerning the inheritance of traits and the influence that one's surroundings has on the development of individuals. As more is learned about the role of genes and how they are passed on to offspring, scientists are better able to distinguish between **acquired traits**, which come from the environment and are not passed on to offspring, and **inherited traits**, which are the result of genes.

In Lesson 2.2 you will apply Mendel's work to making predictions about the genetic inheritance of single traits. Using genetic diagrams, you will analyze the probability of offspring inheriting particular traits. Through the study of autosomal and sex-linked patterns of inheritance, you will also learn why some diseases and characteristics are present in a particular gender more than they are in the other gender.

It is important to keep in mind that the study of human genetics is much more complicated than the introductory concepts taught in Science 30. The proper analysis of human genetic traits requires years of study and training. It follows that you must be cautious when it comes to drawing conclusions from an analysis of genetic traits based solely upon the information presented in this course.

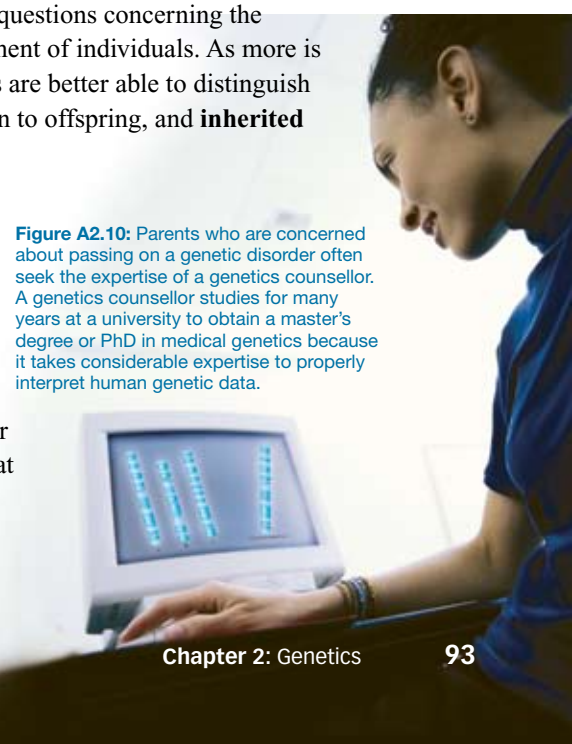


Figure A2.10: Parents who are concerned about passing on a genetic disorder often seek the expertise of a genetics counsellor. A genetics counsellor studies for many years at a university to obtain a master's degree or PhD in medical genetics because it takes considerable expertise to properly interpret human genetic data.

Practice

11. List five traits you may have inherited from your parents.

12. State some acquired traits that cannot be passed on to offspring.

13. Look at the family in the photograph.

The mother and father have different eye colours, hair colours, hair types, and skin colours. Their little girls inherited some characteristics from each parent. Identify which characteristics the girls may have inherited from each parent.

Alleles

At the beginning of Chapter 2 you looked at the ability to roll your tongue. You observed that there are two distinctive traits—some people can roll their tongues and others cannot. The differences in these traits can be traced to alternate forms of a specific gene. These alternate forms of genes are called **alleles**. In the case of tongue rolling, there are two alleles that can produce the two possible traits. One particular allele provides the genetic instructions that create the tongue-rolling trait; so if you do not possess that allele, you will not have the trait.

Individuals possess two alleles for every trait located at specific sites on homologous chromosomes. Since homologues separate during meiosis, only one of these alleles is passed onto each gamete. The two alleles that you inherit are contained in the particular sperm cell and egg cell that joined during fertilization. Therefore, when you think about inheritance, you also have to think about probability. In other words, which one allele of the two possibilities will be in each of the particular gametes that join to form an individual?

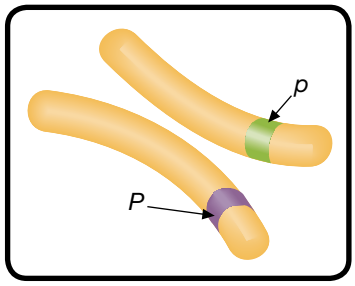


Figure A2.11: Two different alleles are found on a pair of homologous chromosomes.

Dominant Versus Recessive

Gregor Mendel's studies of pea-plant traits that appear in two distinct forms can also be used to help understand how many traits, like tongue rolling, are expressed and passed on. In Mendel's experiments with pea-flower colour, he found that crossing a white-flowered plant with a purple-flowered plant resulted in all offspring producing purple flowers. This means that if an offspring receives a gene in the form of the purple allele from a parent, the purple colour will be produced even if the allele received from the other parent is the white gene. How can this be explained?

In genetic crosses between two individuals, each gene being studied is assigned a letter. Genes can be assigned any form of symbol, but using letters creates a way to easily represent the related, yet different, alleles.

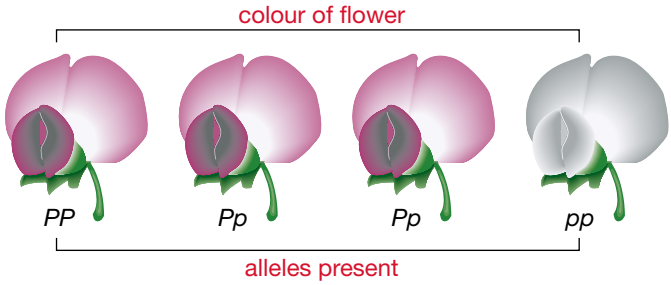


Figure A2.12: Pea plants with purple flowers have at least one copy of the dominante allele, *P*. Pea plants with white flowers have two copies of the recessive allele, *p*.

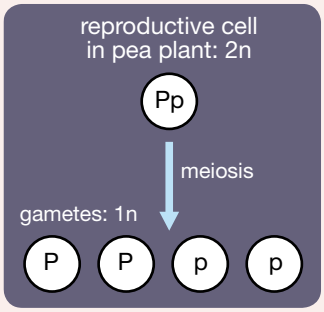
Mendel proposed a theory to describe the occurrence of traits that he observed in his pea-plant studies. Since the purple flower colour trait is dominant to the white trait, Mendel stated that the purple flower colour is a **dominant** trait in pea plants. Dominant traits are caused by dominant alleles. If an offspring receives even one dominant allele, the dominant trait will appear in that offspring. He said that the trait that is masked and not expressed when the dominant allele is present is called the **recessive** trait. Recessive traits are only expressed when an offspring receives two copies of the recessive allele. A white-flowered pea plant has two recessive alleles for flower colour.

The ability to roll your tongue is a dominant trait produced by the dominant allele, which you inherited from at least one of your parents. If you cannot roll your tongue, it means that you have two copies of the recessive allele.

- **allele:** an alternative form of a gene responsible for a trait
- **dominant:** referring to a dominant allele that overpowers a recessive allele—an individual only needs one dominant allele for the dominant trait to be expressed
- **recessive:** referring to a recessive allele that is not expressed when the dominant allele is present—two recessive alleles need to be present for the recessive trait to be expressed in an individual

Example Problem 2.1

At a specific location on a particular chromosome within a pea plant is the gene that determines the flower colour. Since the pea plant has two copies of each chromosome, there are two copies of the gene for flower colour. However, the gene for flower colour located on each chromosome copy may not be identical. For example, one allele, *P*, codes for purple flowers, while the other allele, *p*, codes for white flowers. A reproductive cell within a pea plant can produce four gametes so that two of the gametes have the allele *P*, while the other two have the allele *p*.



Suppose two pea plants each have the allele *P* that codes for purple flowers and the allele *p* that codes for white flowers. Let these two plants be the parent generation.

a. Determine the colour of the flowers in each of the parent pea plants.

b. Set up a chart to show all the possible outcomes of fertilizing a gamete from one plant with a gamete from the other.

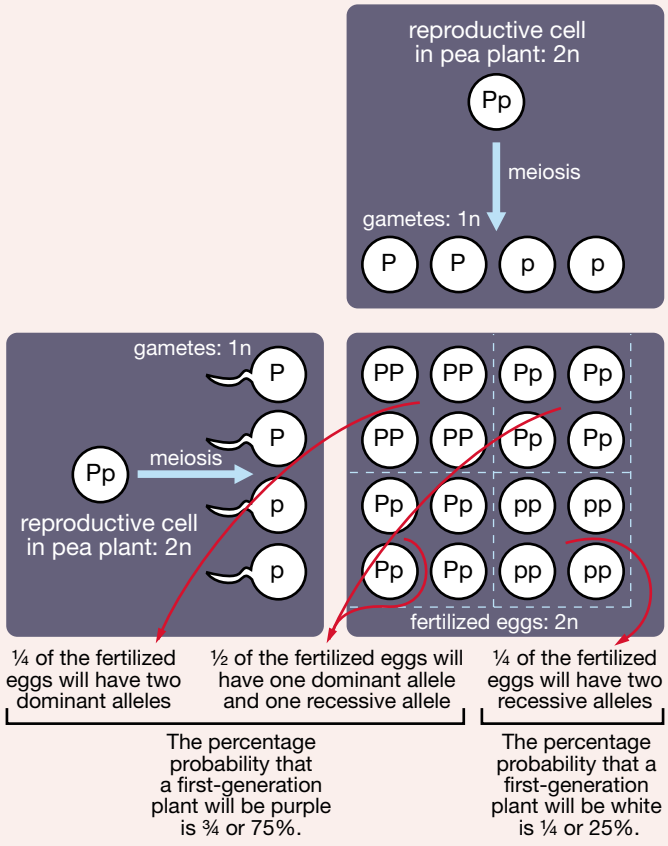
c. Use the chart you developed in b. to determine the percentage probability that a plant in the first generation of offspring will have white flowers.

d. Suppose the two parent plants produced twelve plants in the first generation of offspring. How many of these plants would you expect to have white flowers? Suggest some reasons why the number of plants with white flowers could add up to a different number.

Solution

a. Each of the parent pea plants will produce purple flowers, since the dominant allele, *P*, for purple flowers masks the recessive allele, *p*, for white flowers.

b.



c. The two parent plants each produce four gametes, combining for a total of sixteen possible offspring. Of these offspring, four have the two recessive alleles to produce white flowers. Since four of the sixteen possible outcomes produce white flowers, the percentage probability that a plant in the first generation will have white flowers is 1/4 or 25%.

d. Since the percentage probability of an offspring in the first generation having white flowers is 1/4, then it is most likely that three plants (1/4 of the twelve offspring) will have white flowers. This value assumes that all the gametes are produced and fertilized in the exact proportions predicted by the chart. In reality, some of the sperm may not fertilize an egg and some eggs may not be fertilized by any sperm.

Punnett Squares

As shown in Example Problem 2.1, if the alleles for an inherited trait are known, it is possible to predict the probability of the offspring having a particular genetic make-up or **genotype**.

But the method shown is lengthy and quite repetitious. Note that even though four gametes were produced by each parent plant, there were really only two possibilities: a gamete will either have the dominant allele or the recessive allele.
A streamlined version of this process uses a more concise chart called a **Punnett square**.

As you'll see in Example Problem 2.2, the Punnett square method is very efficient.

How to Use a Punnett Square

step 1: Draw a square and then label each row and column with the alleles of each gamete.

step 2: Fill in the square with the offspring genotype.

step 3: Determine the fraction of the offspring with each genotype. This fraction is the same as the probability of an individual offspring possessing a particular genotype.

genotype: a description of the alleles that an individual possesses
This is communicated by using letters to represent the different allele versions.

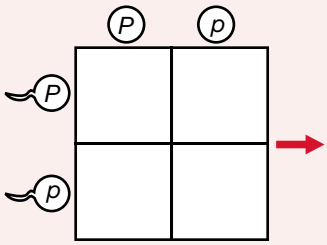
Punnett square: a table that uses the alleles of the parents to indicate all possible outcomes resulting from gamete fertilization

Example Problem 2.2

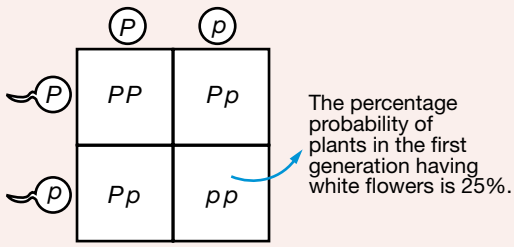
Two pea plants in the parent generation each contain the dominant allele *P* that codes for purple flowers and the recessive allele *p* that codes for white flowers. Use a Punnett square to predict the percentage probability that a plant in the first generation of offspring will have white flowers.

Solution

step 1:



step 2:



Practice

Use the following information to answer questions 14 and 15.

In human beings, the ability to roll one's tongue is dominant over non-tongue rolling. As you solve the following questions, use *R* to represent the dominant allele for tongue rolling and *r* to represent the recessive allele for non-tongue rolling.

14. Two parents each possess the dominant allele *R* and the recessive allele *r*.

Use a Punnett square to determine the percentage probability that their offspring will be able to roll their tongues.

15. One parent possesses the dominant allele *R* and the recessive allele *r*. The other parent possesses two copies of the recessive allele *r*.

a. Are both of these parents able to roll their tongues?

b. Use a Punnett square to determine the percentage probability that their offspring will be able to roll their tongues.

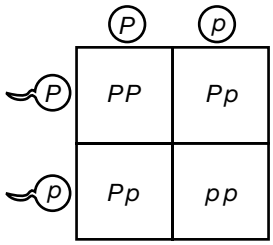
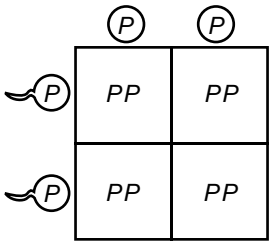
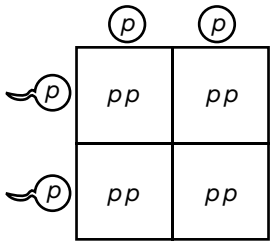
Homozygous and Heterozygous

Punnett squares are a powerful tool to show the outcomes of several types of crosses. When an organism has two copies of the same alleles that are either both dominant or both recessive, the organism is called **homozygous**. In the case of pea plants, the purple flowers are homozygous for the dominant condition and the white flowers are homozygous for the recessive condition. When an organism possesses one dominant allele and one recessive allele for a trait, it is said to be **heterozygous** for that trait.

The following table shows some of these possibilities.

- **homozygous:** referring to an organism that has two copies of the same allele for a given trait—*pp* or *PP*
- **heterozygous:** referring to an organism that has a dominant allele and a recessive allele for a given trait—*Pp*

USING PUNNETT SQUARES TO PREDICT THE OUTCOMES OF GENETIC CROSSES

Heterozygous Parents	Homozygous Dominant Parents	Homozygous Recessive Parents
Two pea plants, each with one dominant allele, *P*, and one recessive allele, *p*, are crossed.	Two pea plants, each with two copies of the same dominant allele, *P*, are crossed.	Two pea plants, each with two copies of the same recessive allele, *p*, are crossed.
		
Possible outcomes of first generation offspring: • 1/2 of the offspring are **heterozygous** • 1/4 of the offspring are **homozygous** for the dominant allele *P* • 1/4 of the offspring are **homozygous** for the recessive allele *p*	Possible outcomes of first generation offspring: • all the offspring are **homozygous** for the dominant allele *P* • all the offspring are true breeding for purple flowers	Possible outcomes of first generation offspring: • all the offspring are **homozygous** for the recessive allele *p* • all the offspring are true breeding for white flowers

The left column summarizes the work in Example Problems 2.1 and 2.2. Each time Mendel set up a heterozygous cross, he noted that close to 1/4 of the offspring demonstrated the recessive trait and about 3/4 of the offspring demonstrated the dominant trait. Remember, the fractions that result from Punnett squares only indicate the probability of that characteristic appearing with each cross.

Practice

In people, the ability to roll a tongue is dominant over non-tongue rolling. As you solve the following problems, use *R* to represent the dominant allele for tongue rolling and *r* to represent the recessive allele for non-tongue rolling.

16. Use letters to describe the genotype of each following individual.

- **a.** a homozygous tongue roller
- **b.** a heterozygous tongue roller
- **c.** a homozygous non-tongue roller

17. **a.** Draw a Punnett square for the cross of a homozygous tongue roller with a homozygous non-tongue roller.

- **b.** State the likely percentage probability that the offspring will be able to roll their tongues.
- **c.** State the likely percentage probability that the offspring will not be able to roll their tongues.
- **d.** State the likely percentage probability that the offspring will be able to roll their tongues but will also carry the recessive non-tongue rolling gene.

Genotype Versus Phenotype

Since dominant traits are expressed and recessive traits are masked, if a dominant allele is present it is easy to determine whether someone possesses at least one dominant allele. If you can roll your tongue, it means that you have a dominant allele for tongue rolling. The physical expression of the alleles that you possess is called the **phenotype**. The phenotype for the tongue-rolling trait would either be *tongue roller* or *non-tongue roller*. An organism's genotype is a description of the alleles that it possesses. The genotype for tongue rolling could be homozygous dominant (*RR*) heterozygous (*Rr*) or homozygous recessive (*rr*). It should be noted that a person with the genotype *RR* and the genotype *Rr* both have the same phenotype since they can both roll their tongues, even though their genotypes are different.

phenotype: the physical and physiological traits of an organism

Practice

Use the following information to answer questions 18 to 20.

Nectarines and peaches are genetic variations of the same fruit. The fuzzy skin of a peach is produced by a dominant allele, *N*, and the smooth skin of a nectarine is produced by a recessive allele, *n*.

18. State whether the skin phenotype of the following individuals is fuzzy or smooth.

- **a.** *NN*
- **b.** *Nn*
- **c.** *nn*

19. State the likely genotype of each example.

- **a.** a smooth-skinned nectarine
- **b.** a fuzzy-skinned peach bred from a cross between a peach-producing tree and a nectarine-producing tree
- **c.** a fuzzy-skinned peach produced from a long line of peach-producing trees

20.
- **a.** Draw a Punnett square for a cross between a heterozygous peach and a homozygous nectarine.
- **b.** Use your answer from question 20.a. to determine the probability of this cross producing the genotypes *NN*, *Nn*, and *nn*.
- **c.** Use your answer from question 20.a. to determine from this cross the probability of offspring produced with the smooth-skinned nectarine phenotype and the probability of offspring produced with the fuzzy-skinned peach phenotype.
- **d.** Determine what percentage of the offspring will carry the allele for smooth skin.
- **e.** Is it possible for a nectarine to be heterozygous for the skin-type trait? Explain.

21. The ability to taste the chemical phenylthiocarbamide (PTC) is dominant over the inability to taste the chemical. Observe the following Punnett square from a cross between a male PTC taster and a female PTC non-taster (*TT* × *tt*).

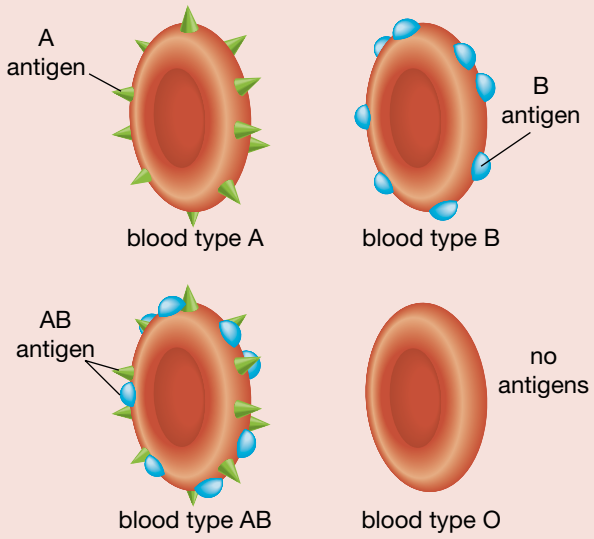
	t	*t*
T	*T t*	*T t*
T	*T t*	*T t*

- **a.** Describe the PTC tasting genotype of the offspring.
- **b.** Describe the PTC tasting phenotype of the offspring.

22. In people, curly hair is dominant over straight hair. A homozygous, curly haired man (*CC*) is about to have a child with a homozygous, straight-haired woman (*cc*).

- **a.** Draw the Punnett square for this cross.
- **b.** Determine the probability that their child will have curly hair.
- **c.** Re-examine the photo of the family on pages 76 and 77. Use a Punnett square to suggest an explanation for the child's straight hair.

Other Mechanisms of Inheritance



Note: Antigens are not drawn to scale.

Not all traits are controlled by one gene or have only two alleles for a gene. Hair and eye colour do not appear in only two forms because they are controlled by more than one pair of genes. Blood type is an example of a trait with more than two possible alleles. There are three forms or alleles of the blood-type gene represented by the letters *A*, *B*, and *O*. These three alleles can produce four phenotypes. The different forms of *A* and *B* produce a modified surface protein on red blood cells that give the cell its unique phenotype. In this case, the phenotype is observed in terms of the type and presence of antigens on the surface of the blood cell. The *O* allele is a recessive form of the gene that does not produce a modified surface protein and can be masked by the *A* and *B* alleles. To express the *O* blood type, an individual must be homozygous for the *O* allele. Although the *A* and *B* alleles are both dominant over the *O* allele, *A* and *B* are different modifications of the surface and do not mask one another. Since neither state *A* nor *B* is dominant over the other, they are said to exhibit codominance, which is a condition where both allele products are expressed at the same time. This results in the *AB* phenotype.

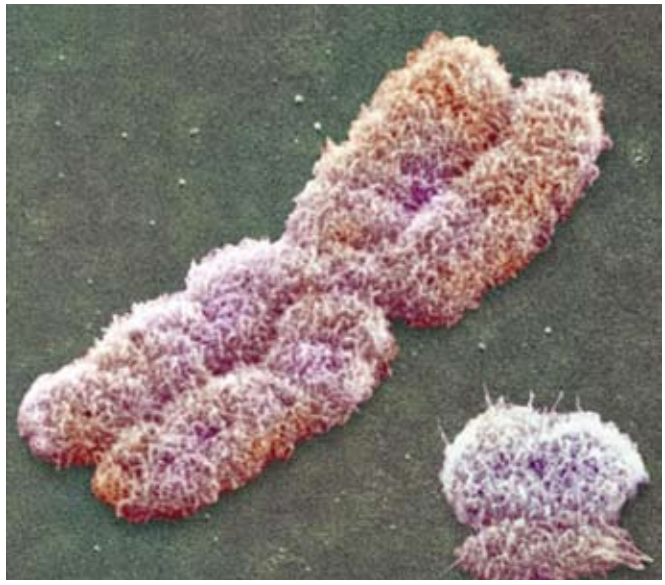
If you know your blood type, include this information in your health file.

Table of Blood Types

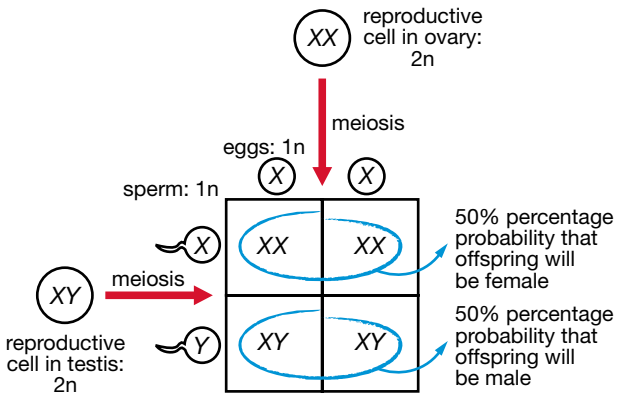
Genotype	Phenotype
OO	blood type *O*
AA or *AO*	blood type *A*
BB or *BO*	blood type *B*
AB	blood type *AB*

Boy or Girl? Determining Gender

In Lesson 2.1 you looked at the different chromosomes present in a cell nucleus. If you compare the chromosome pairs of human males and females, you will find that 22 of the 23 pairs look the same. The one major difference between the chromosomes of a male and a female is that females possess two *X* chromosomes, whereas males possess one *X* chromosome and one *Y* chromosome. *X* and *Y* chromosome inheritance determines the gender of an offspring. You can use a Punnett square to illustrate how gender is determined by an offspring's inheritance of the *X* and *Y* sex chromosomes.



Since the female, or mother, only has *X* chromosomes to give, all her eggs produced during meiosis normally contain a single *X* chromosome. The male, or father, has either an *X* chromosome or a *Y* chromosome to give, so his sperm will normally have either an *X* or a *Y*. When you make a Punnett square, you can see that there is always a 50% percentage probability of the gametes uniting to become a boy and a 50% chance of the gametes combining to become a girl. These percentages are probabilities, like the 50% chance of getting heads when flipping a coin. It does not mean that children will be born boy, girl, boy, girl . . . .



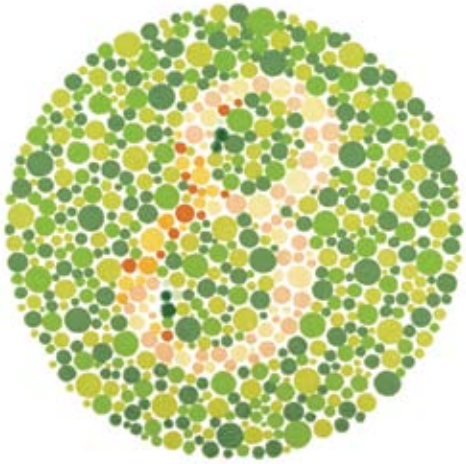
Traits Associated with the *X* Chromosome

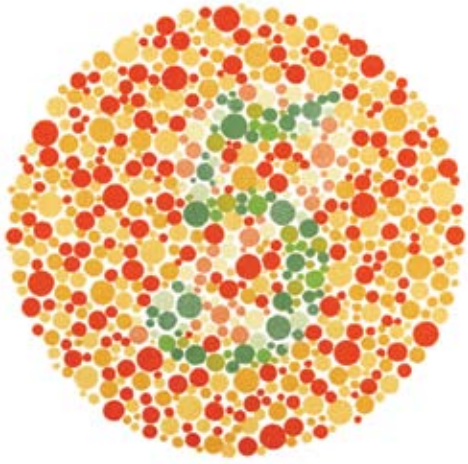
The *X* and *Y* chromosomes that determine gender also carry other genes that do not determine the sexual characteristics of the individual. Genes like these are said to be responsible for **sex-linked inheritance**. Genes carried on the other 22 pairs of chromosomes are said to be responsible for **autosomal inheritance**.

- **sex-linked inheritance:** traits not directly related to primary or secondary sexual characteristics that are coded by the genes located on the sex chromosomes
- **autosomal inheritance:** traits controlled by genes found on the 22 pairs of autosomal chromosomes

Colour-Blindness

The term *colour-blindness* is used to refer to the inability to perceive differences between some or all colours readily recognized by people with full-colour vision. There are many types of colour-blindness. Some are caused by damage to the eyes or the optic nerves, but most are hereditary. In this chapter, colour-blindness will refer to red-green colour-blindness. People with red-green colour-blindness would be unable to identify the 8 or 5 within the following circles. This is one of the most common types of colour-blindness, and it is caused by sex-linked inheritance.





DID YOU KNOW?

Colour-blindness is often assumed to be a disability because persons with colour-blindness are unable to detect colours to the same degree as persons without colour-blindness. However, whether a condition is a disability, an inconvenience, or an ability depends very much upon the specific environment the person is in.

In a hunting or a military environment, people with colour-blindness have an advantage when it comes to seeing objects against confusing backgrounds. In this case, colour-blindness should be referred to as *special counter-camouflage ability*.

Clearly, the context provided by the environment plays a key role in determining whether a condition is also a disability.

A Punnett square can be used to make predictions about sex-linked traits. The traits are represented as uppercase and lowercase letters for dominant and recessive alleles, like they are with autosomal traits. The only difference between autosomal and sex-linked Punnett squares is that the letters used to represent the sex-linked traits for alleles are written as superscripts above the chromosome on which they are carried. Let *N* represent the allele for full-colour vision and let *n* represent a recessive allele that produces the condition of colour-blindness.

Because colour-blindness is an *X* chromosome, sex-linked trait, the allele is not carried on the *Y* chromosome, and the possible genotypes are X^N and X^n. A complete male geneotype for a colour-blind man would be represented as X^nY and a man with full-colour vision would be represented as X^NY. A female with full-colour vision who carries the recessive allele for colour-blindness would be written as X^NX^n and a colour-blind female would be written as X^nX^n.

Example Problem 2.3

A homozygous woman who has two alleles for full-colour vision has children with a colour-blind man.

a. Describe the genotype of each parent.

b. Build a Punnett square to predict the possible genotypes of their children.

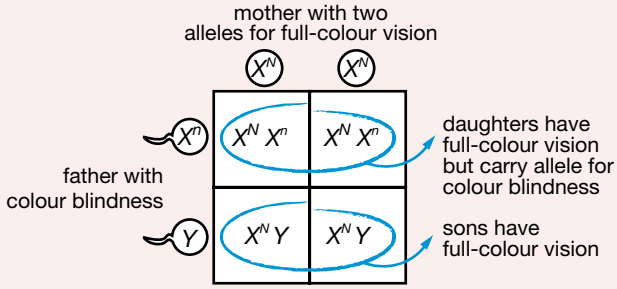
c. Use the Punnett square to explain why the sons have full-colour vision even though their father is colour-blind.

d. Use the Punnett square to explain why the daughters are carriers for the colour-blind allele, even though they have full-colour vision.

Solution

a. The mother's genotype would be X^NX^N, and the father's genotype would be X^nY.

b.



c. The sons are male because they inherited a *Y* chromosome from their father and an *X* chromosome from their mother. Since the *X* chromosome is the location of the allele for colour-blindness, and since the sons inherited this allele from their homozygous dominant mother, the sons have full-colour vision.

d. The daughters are female because they have inherited an *X* chromosome from their mother and an *X* chromosome from their father. Since the *X* chromosome is the location of the allele for colour-blindness, and since the *X* chromosome from their father contains this allele, each daughter is a carrier of the allele for colour-blindness. However, the daughters are not colour-blind themselves because they inherited an *X* chromosome for full-colour vision from their mother.

Practice

23. a. Draw a sex-linked Punnett square for the cross between a man with full colour vision and a woman with full-colour vision who is a female carrier of the recessive colour-blind allele.

b. Determine the percentage probability of their sons being colour-blind.

c. Determine the percentage probability that their daughters will be carriers of the recessive colour-blind allele even though the daughters have full-colour vision.

24. a. Draw a sex-linked Punnett square for the cross between a colour-blind man and a woman with full-colour vision who is a female carrier of the recessive colour-blind allele.

b. Determine the percentage probability of their sons being colour-blind.

c. Determine the percentage probability of their daughters being colour-blind.

d. Determine the percentage probability that their daughters have full-colour vision but will be carriers of the recessive colour-blind allele.

25. An expectant father who is colour-blind is afraid that his soon-to-be born son will also be colour-blind. Explain to this expectant father why it is best to look at the mother's side of the family for an indication about whether their son will become colour-blind.

26. Determine which gender would be most affected if a trait were found only on a gene from the *Y* chromosome.

Investigation

Investigating Dominant and Recessive Human Traits

Background Information

Several human physical traits are both distinctive and easily observed. Like tongue rolling, they are caused by the presence of either dominant or recessive alleles inherited from parents.

Science Skills

- ✓ Performing and Recording
- ✓ Analyzing and Interpreting

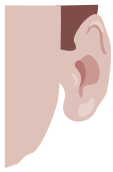
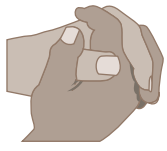
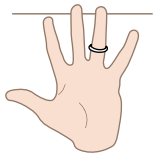
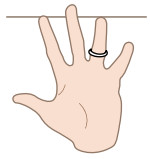
Purpose

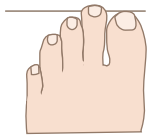
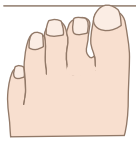
You will survey the members of your class to obtain data on the presence of dominant and recessive alleles.

Procedure

Survey each person to discover if he or she has a dominant or recessive trait for the following eight traits. Record your findings in a table.

DOMINANT OR RECESSIVE TRAITS

Trait	Dominant Phenotype	Recessive Phenotype
(1) hair type	curly hair (genotype *CC* or *Cc*) 	straight hair (genotype *cc*) 
(2) hairline	widow's peak (genotype *WW* or *Ww*) 	straight hairline (genotype *ww*) 
(3) ear lobes	free floating (genotype *EE* or *Ee*) 	attached (genotype *ee*) 
(4) left-handed or right-handed	right-handed (genotype *RR* or *Rr*) 	left-handed (genotype *rr*) 
(5) thumb position with hand folding	left thumb over right thumb (genotype *TT* or *Tt*) 	right thumb over left thumb (genotype *tt*) 
(6) finger length	ring finger longer than index finger (genotype *FF* or *Ff*) 	index finger longer than ring finger (genotype *ff*) 

(7) thumb curvature	"hitchhiker's thumb" (genotype *DD* or *Dd*) 	straight thumb (genotype *dd*) 
(8) second toe	second toe longer than big toe (genotype *GG* or *Gg*) 	second toe shorter than big toe (genotype *gg*) 

Analysis

1. Use your recorded class data to draw a bar graph with a dominant bar and a recessive bar for each trait.
2. Observe the number of traits where the dominant phenotype is greater than the recessive phenotype. Identify a reason for the higher frequency of certain traits in the population by using a Punnett square.
3. List your own phenotype for all eight traits.
4. Explain why it is difficult to accurately list your genotype for all eight traits.
5. Describe the phenotype of the individual who has the following genotype based on the letter used for the traits in the "Dominant or Recessive Traits" table: *CC*, *Ww*, *EE*, *rr*, *Tt*, *Ff*, *DD*, *gg*.
6. Perform the following crosses for the traits studied in the table by preparing a Punnett square for each noted cross. State the predicted genotype ratios and phenotype ratios of the offspring for each cross noted.
   - **a.** An individual homozygous for attached ear lobes has a child with a heterozygous free-floating, ear-lobed person.
   - **b.** *GG* × *gg*
   - **c.** A straight-haired individual has a child with another straight-haired individual.

2.2 Summary

Traits such as hair colour and eye colour passed on from your parents are called inherited traits. Traits learned or gained from the results of experiences, such as languages and injuries, are called acquired traits.

The forms of a gene are called alleles and can be either dominant or recessive. Dominant alleles are expressed or they mask the presence of recessive alleles. The effect of recessive alleles is observed only when an individual possesses two recessive copies of the allele for that particular trait.

Punnett squares are one way to predict the probability of inheriting genetic traits. Letters are used to represent genes in a Punnett square cross—upper-case letters are used to represent dominant alleles and lower-case letters are used to represent recessive alleles. When the two copies of an allele are either both dominant or both recessive, it is called a homozygous condition (e.g., *PP* or *pp*); and when the two genes are a different allele form, it is called heterozygous (e.g., *Pp*).

The expression of genes as observable characteristics is called a phenotype and is determined by the alleles present in an individual's genotype.

Punnett squares can also be used to predict the gender of offspring. Some genes that do not directly relate to sexual characteristics are found on sex chromosomes. These genes are said to be sex-linked rather than autosomal. Colour-blindness is an example of a trait that affects men more often than women.

2.2 Questions

Knowledge

1. Jim has dark curly hair, brown eyes, and a large scar on his cheek. As a child, he regularly practised the piano and became a gifted pianist. He is a skilled downhill skier and loves all winter sports. From this description, list Jim's genetically inherited traits and the traits that he has acquired.
2. A genotype for the fur-colour trait in mice is abbreviated as *Mm*.

   a. State the dominant allele in the genotype.
   b. State the recessive allele in the genotype.
   c. Is this individual described as homozygous or heterozygous?
   d. If black fur is dominant over white fur in mice, state the phenotype of the mouse with the genotype *Mm*.
3. In cats, the gene that causes the ginger- or orange-fur colour is a sex-linked trait carried on the *X* chromosome. The ginger colour (*G*) is dominant to the black colour (*g*).

   a. Write the genotype for a ginger male cat.
   b. Describe the phenotype of a cat with the genotype X^gY.
   c. Describe the phenotype of a cat with the genotype X^GX^G.

Applying Concepts

4. A family has three girls and is expecting a fourth child. What is the probability that the fourth child will be a boy?
5. Explain the difference between autosomal inheritance and sex-linked inheritance.
6. In garden peas, the yellow-seed colour is an autosomal dominant trait over the green-seed colour.

   a. Choose letters to represent the dominant and recessive alleles for this trait. Write the genotypes for a pea plant that is homozygous for yellow, homozygous for green, and heterozygous for yellow.
   b. Draw a Punnett square for a cross between a homozygous yellow-seeded pea plant and a homozygous green-seeded pea plant. State the predicted genotypes and phenotypes of the offspring.
   c. Draw a Punnett square for a cross between two of the offspring produced in question 6.b. State the predicted genotypes and phenotypes of the offspring.
7. The gene for eye colour in fruit flies is located on the *X* chromosome. The allele for the dominant red-eye colour could be represented by the allele X^R, while the allele for the recessive white-eye colour could be represented by the allele X^r.
   a. Draw a sex-linked Punnett square for a male with red eyes who breeds with a female with white eyes.
   b. Determine the percentage probability that the male offspring will have white eyes.
   c. Determine the percentage probability that the daughters will have white eyes.
   d. Determine the percentage probability that the female offspring are carriers of the recessive white-eyed allele.

2.3 DNA

Have you ever listened to music at an outdoor concert on a warm summer's evening? If you have, can you remember the particular songs or pieces that were played? Depending upon the type of music and the performers, the musicians sometimes memorize the music. In other cases they use sheet music. On one level, a performer who is reading the sheet music is translating the information from the page into a tune or a song. But lovers of music would argue that there is much more to performing than playing the notes in the right order—a good musician is an artist.

To someone who doesn't read music, the symbols used for notes look like simple shapes on a page in random order. The information on a deoxyribonucleic acid (DNA) molecule might also seem like a random arrangement of chemical units, but to the cells in your body this arrangement is a meaningful set of instructions for making essential products. Just like a huge variety of songs can be written on sheet music from a limited number of musical notes, a huge variety of instructions can be encoded on a DNA molecule from a limited number of chemical units. While music is the product that can be produced by playing the notes on sheet music, proteins are the product that can be produced by translating the code on a DNA molecule. These proteins make life possible by forming structural and regulatory molecules within cells.

Understanding how DNA works is a bit like understanding sheet music. First, you need to understand how the symbols are used to represent the sound that each note makes and the rules used in writing music. Once you understand the language of music, then you can look at how the notes are put together for a particular song and can use an instrument to translate written notes into played notes. Recognizing the symbols used to represent the chemical units of DNA and understanding how the DNA molecule is put together are important skills that allow scientists to read and translate this genetic information into the protein products that make up an organism.

In this lesson you will identify the structure and components of DNA. You will learn how to read and translate elementary sections of DNA code and explain the process of DNA replication. Just as there is more to music than playing notes in the correct order, there is more to a full understanding of DNA than learning how to assemble simple structures. Nevertheless, just as some musical training leads to a greater appreciation of a performing artist's skill, your work with DNA will provide you with a new appreciation of the instructions that made you.

Try This Activity

Extracting DNA from Wheat Germ

Purpose

In this investigation you will extract DNA from raw wheat germ.

Materials

- 1 g of raw wheat germ (not toasted)
- liquid detergent
- 95% ethanol (ethyl alcohol) or 70% isopropyl alcohol
- 2 50-mL test tubes with stoppers
- hot tap water (approximately 50–60°C)
- graduated cylinder
- 2 glass stirring rods
- paper towel
- paper clip

Science Skills

✓ Performing and Recording
✓ Analyzing and Interpreting

Procedure

step 1: Add 20 mL of warm tap water to both test tubes.

step 2: Add 1 g of raw wheat germ to only one of the test tubes. Mix continuously for at least 3 min.

step 3: Add 1 mL of liquid detergent to each test tube, and use a glass stirring rod to stir each gently for 5 min, creating as little foam as possible. Vigorous stirring will break up any DNA into smaller pieces, which are more difficult to see.

step 4: Use a sheet of paper towel to remove any foam produced.

step 5: Tilt the test tubes and slowly pour 14 mL of alcohol down the side of each test tube. The alcohol should form a separate layer on top of the test-tube mixture. Do not stir or mix the two layers.

step 6: Let the test tube sit for a few minutes. Extracted DNA will appear as a white filmy substance and should precipitate and clump together at the boundary of the two layers. If you continue to let it sit, the DNA should float to the top of the alcohol or you can use your glass stirring rod or the loop of a partially straightened paper clip to pull the DNA out of the solution.

Analysis

1. Explain why you prepared a test tube with wheat germ and then—following the identical procedure—another test tube without any wheat germ.
2. Compare the amount of DNA in the test tube with the wheat germ to the test tube without the wheat germ.
3. Do you think it would be possible to use a similar procedure to extract DNA from other materials such as split-green peas, strawberries, or chicken livers? Explain why.

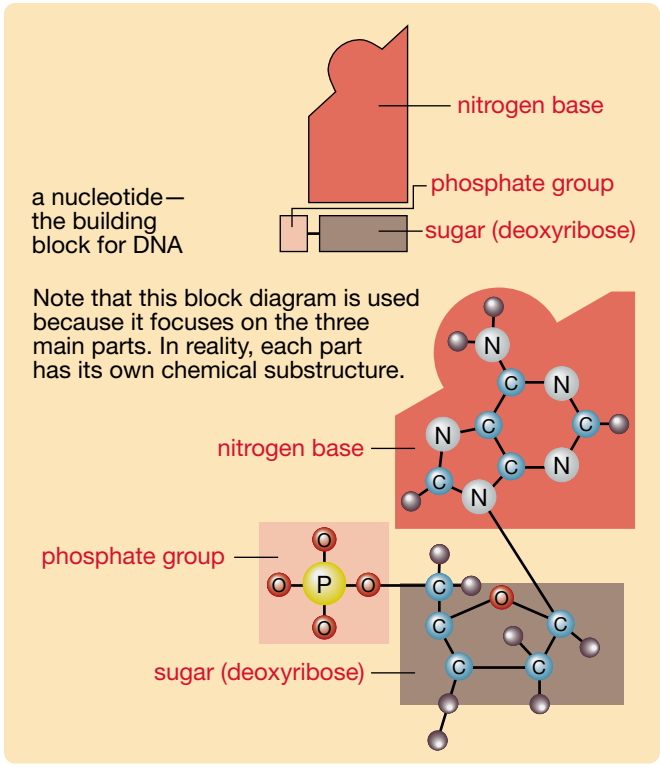
Figure A2.13

The Structure of DNA

The overall shape of a DNA molecule has been compared to a twisted ladder or a spiral staircase. Just as the spiral staircase in Figure A2.13 was assembled from individual wedge-shaped steps, the DNA molecule is composed of chemical units called **nucleotides**. Each nucleotide contains a phosphate molecule, a sugar called deoxyribose, and one of four nitrogen-base molecules.

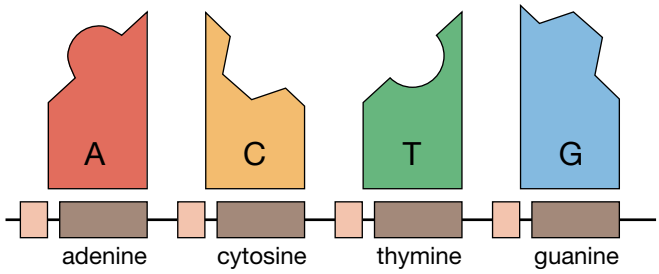
nucleotide: a chemical unit consisting of a phosphate molecule, a deoxyribose sugar molecule, and one of the four nitrogen-base molecules—adenine, cytosine, thymine, or guanine

Two complementary nucleotide chains combine to form DNA.



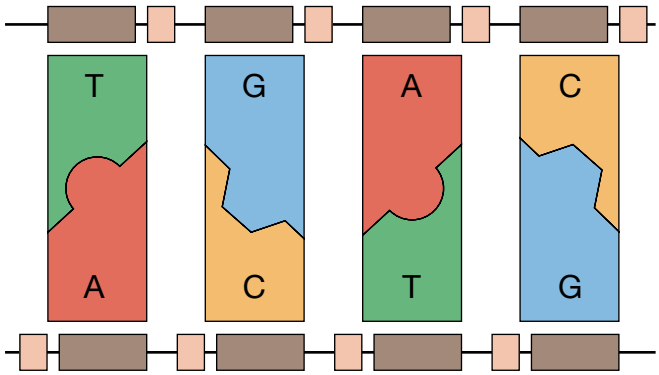
The phosphate and sugar parts of the nucleotide attach to each other to form a repeating chain that makes up the "backbone" of a DNA molecule. The nitrogen base part of the nucleotide sticks out from the sides of this chain. The four nitrogen bases are adenine, cytosine, thymine, and guanine.

The four types of nitrogen bases in nucleotides join to form a single chain—half of a DNA molecule.



These bases are usually abbreviated using the first letter of their name. The long chain of nucleotides can be very long—in this arrangement it makes up half of the DNA molecule. The other half of a DNA molecule is formed from another long chain of nucleotides that attaches to the first strand of nucleotides by hydrogen bonds between the nitrogen bases on opposite strands.

DNA consists of two long chains of nucleotides connected by complementary nitrogen base pairs.

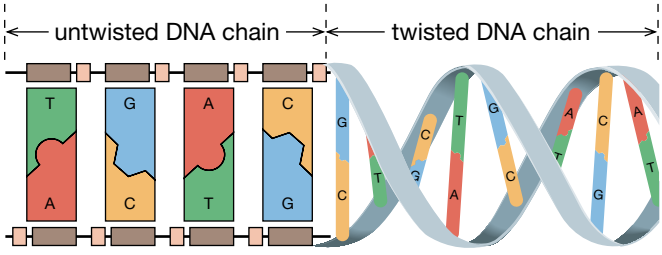


base pair: the two nucleotides connected on opposite sides of complementary strands of the DNA molecule

Complementary base pairings for DNA are adenine with thymine and cytosine with guanine.

The four possible bases have a unique chemical structure that allows them to bond only with one other base. When the two bases are bonded together, they are called a **base pair**. Adenine can only bond to thymine and vice versa (A-T or T-A) and cytosine can only bond to guanine and vice versa (C-G or G-C). As a result of the specific bonding between bases, the DNA molecule is comprised of two long chains of nucleotides with bases of one chain paired up with another chain containing complementary bases. For example, if the base pairs on one side of the molecule are ACTGTTA, then the other side of that section of DNA has the complementary base pairs of TGACAAT. The two paired strands form a structure that looks like a twisted ladder, with the base pairs acting like the rungs of the ladder and the sugar and phosphate molecules acting as the ladder's side. The distinctive coiled shape of DNA, which is similar to the appearance of a spiral staircase or a twisted phone cord, is called a double helix.

The double-helix shape of DNA is made as the two interlocking chains of nucleotides are twisted.



Utilizing Technology

Building a DNA Segment

Science Skills

✓ Analyzing and Interpreting

Purpose

You will build a short segment of the DNA strand by matching up the nitrogen bases in the nucleotides.

Procedure

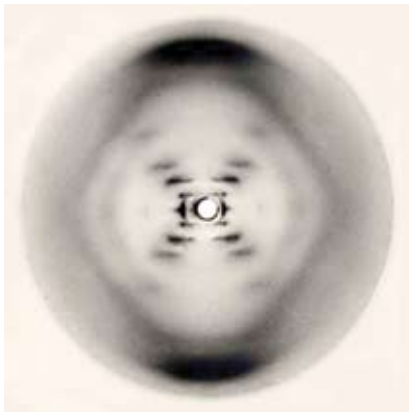
Locate the applet "Building a DNA Molecule" on the Science 30 Textbook CD. Follow the instructions.

Discovering the Structure of the DNA Molecule

The discovery of the three-dimensional structure of the DNA molecule is credited to James Watson and Francis Crick. Their description of DNA's structure was only possible by incorporating the findings of other scientists whose experiments produced unexplained results.

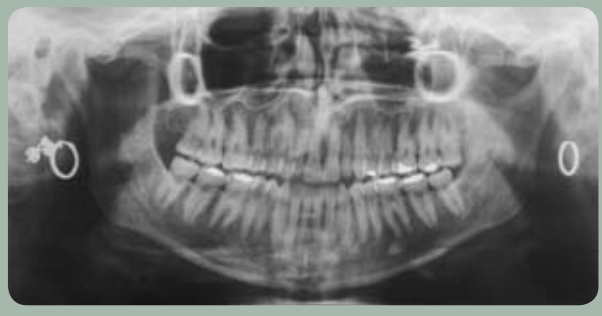
For example, Erwin Chargaff discovered a one-to-one ratio between the bases of adenine and thymine and between the bases of cytosine and guanine. Chargaff could not explain this ratio found in all DNA samples analyzed.

When Rosalind Franklin fired a beam of X-rays at a hair-like thread of DNA, the X-rays changed direction as they encountered the delicate DNA structures. After passing through the DNA, the redirected X-rays created a distinct pattern—coincidentally, the letter X—on photographic film on the other side. Franklin interpreted this photo to mean that the X-rays had encountered a molecule shaped like a helix. She had also discovered that the phosphate part of the molecule was on its outside. Franklin's supervisor Maurice Wilkins, who began the X-ray research, showed her X-ray picture and the results of her work to Watson and Crick without her knowledge or permission. Watson and Crick were able to use Franklin's findings to help assemble a working structure. Franklin died of ovarian cancer in 1958, four years before Watson, Crick, and Wilkins jointly received the Nobel Prize in medicine or physiology for their work on the structure of DNA. The Nobel Prize is awarded only to living scientists.



Science Links

In Franklin's work, the X-rays passed through the DNA but changed direction in the process. This left an interesting pattern on photographic film. When you go to a dentist for teeth X-rays, high energy X-rays are used to create a shadow image of your teeth. Denser areas—corresponding to fillings and jewellery—create shadows on the photographic film placed in your mouth. These shadow areas appear white. You'll learn more about X-rays and how they are used in Unit C.



Practice

27. The nucleotides are the building blocks of the DNA molecule.

a. Sketch the nucleotide that has thymine as its nitrogen base, and label the three distinct parts of the nucleotide.

b. The DNA molecule has been described as a twisted ladder. Add labels to your sketch from question 27.a. to indicate which part(s) of the nucleotide will form the rungs of the twisted ladder and which part(s) will form the long sides.

28. Four types of nucleotides can be identified by the individual nitrogen bases or their abbreviations—adenine (A), cytosine (C), thymine (T), and guanine (G).

a. Identify the complementary base pairs that form in a DNA molecule.

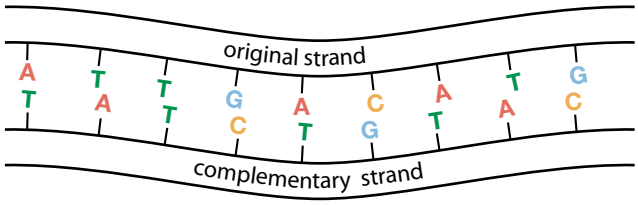
b. State the reason why the nucleotides can pair up only in these combinations.

29. Write the base sequence that makes up the complementary strand for the nucleotide sequence of each provided strand.

a. AAATGTCGCCT **b.** TAGTCTA **c.** GATTGATTCCGGGCTAA

30. A student correctly copied down a nucleotide sequence but made a mistake when writing the complementary strand below it. Identify the mistake in the complementary strand.

There is an error in the complementary strand.

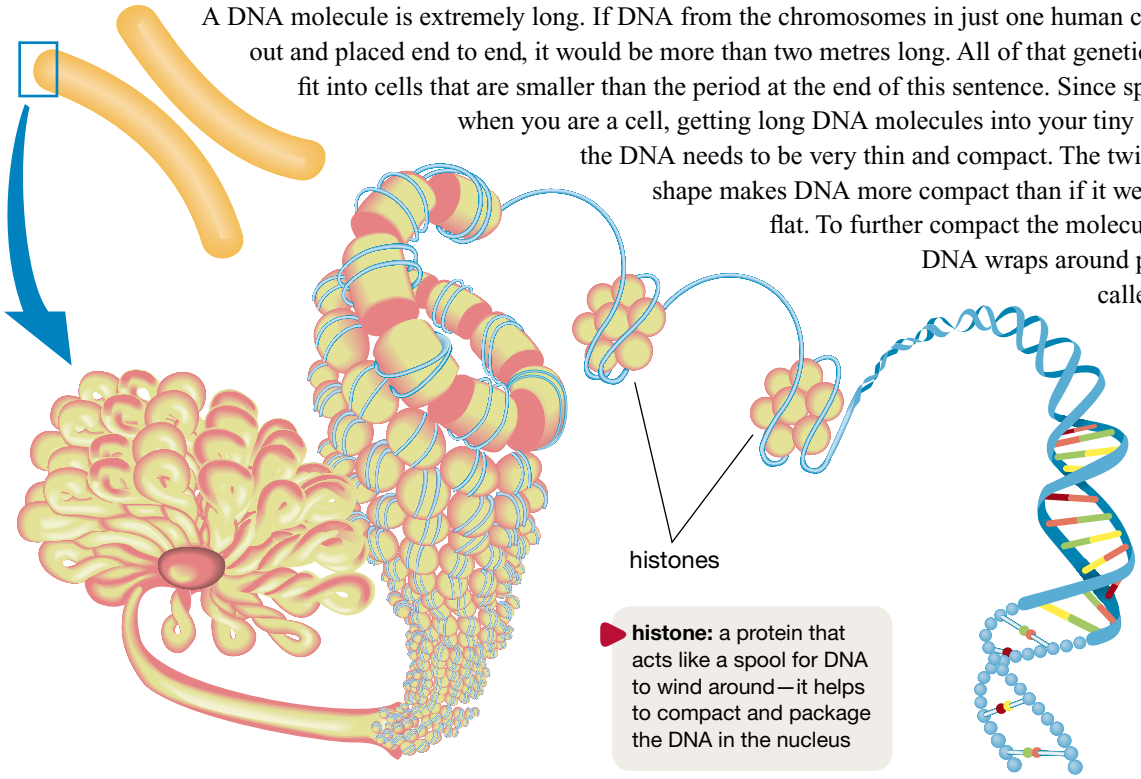


31. Using what you know about DNA structure, account for the findings of Erwin Chargaff.

32. Describe Rosalind Franklin's contribution to the discovery of the DNA molecule's structure.

Histones—Spools for DNA

A DNA molecule is extremely long. If DNA from the chromosomes in just one human cell was stretched out and placed end to end, it would be more than two metres long. All of that genetic material has to fit into cells that are smaller than the period at the end of this sentence. Since space is very limited when you are a cell, getting long DNA molecules into your tiny nucleus means that the DNA needs to be very thin and compact. The twisted double-helix shape makes DNA more compact than if it were uncoiled and flat. To further compact the molecule, the coiled DNA wraps around protein spools called **histones**.



histone: a protein that acts like a spool for DNA to wind around—it helps to compact and package the DNA in the nucleus

In the next investigation you will have an opportunity to explore the advantages of spooling when it comes to packaging long strings of material.

Investigation

Packaging DNA

Purpose

You will perform a simulation that relates how DNA is packed to fit into the nucleus of a cell, and you will also perform calculations involving DNA length.

Science Skills

- ✓ Performing and Recording
- ✓ Analyzing and Interpreting

Materials (for each group of students)

- one spool of sewing thread
- scissors
- tape measure or metre-stick
- one size '000' or '00' empty gelatin capsule per group—available at health food and supplement stores

Procedure

step 1: Imagine that the small gelatin capsule that your group has been given represents an enlarged nucleus.

step 2: Measure and cut a piece of thread 10-m long from your group's spool. This piece of thread will represent the DNA from one set of chromosomes that has been unraveled, attached end to end, and enlarged five times.

step 3: Your objective is to coil and wrap the thread so that it is compact enough to be inserted into the gelatin capsule and the capsule can be closed.

Analysis

1. Was this activity difficult to perform? Were you able to get the thread into the capsule? Could you have put the thread into the capsule without coiling and wrapping it?
2. **a.** Describe how the method you used to get thread into the capsule is similar to the way DNA is compacted and packaged into the nucleus.
   b. Describe how it is different.
3. Explain some methods or tools that would have made an easier job of getting the thread into the capsule.

Calculations

4. The nucleus of an average cell has a radius of about 5 micrometres or 5.0×10^{-3} mm.
   a. Calculate the volume of a spherical nucleus using the formula for the volume of a sphere: $V = \frac{4}{3}\pi r^3$. Express your answer in mm^3.
   b. Measure both the length and radius of your gelatin capsule in millimetres. Use your measurements to calculate the approximate capsule volume by using the formula for the volume of a cylinder: $V = \pi r^2 L$. Your teacher may provide the dimensions of the capsule from the bottle.
   c. Calculate the ratio of how many times larger the capsule you used for the activity is than an actual human cell nucleus. Do this by dividing capsule volume by nucleus volume.
5. It is often stated that within the nucleus of a typical human cell, the total length of all the DNA would be about 2 m if it were stretched out end to end. Use the following information to confirm this statistic:
   - There are two sets of chromosomes in each nucleus.
   - One set of chromosomes has approximately three billion base pairs (3.0×10^9).
   - One base pair is approximately 0.34 nanometres in length (3.4×10^{-10} m).
6. An adult human body has at least 50 trillion cells (50×10^{12}). Use this information and your answer to question 5 to determine the total length of all DNA in a human body if it were stretched out end to end.
7. The distance from the Sun to Earth is 1.5×10^{11} m. This is called one astronomical unit or A.U. Calculate the ratio of approximately how many times longer the total length of DNA in a human body is relative to the distance between Earth and the Sun.
8. Consider your answers to questions 4.a., 6, and 7. Comment on these values in light of your struggles to pack 10 m of thread into a gelatin capsule.

One of the best ways to understand the process of copying a strand of DNA is to try a simple pencil-and-paper version yourself. This is what you will be doing in the next activity.

Try This Activity

Simulating DNA Replication

Purpose

You will use simple materials to simulate the process of DNA replication.

Science Skills

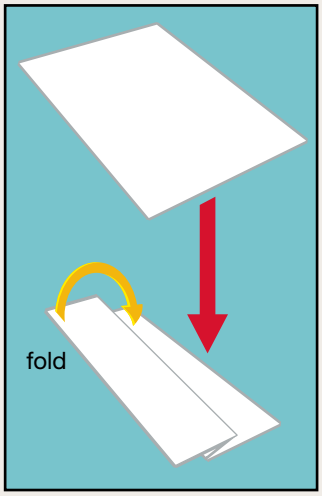
- ✓ Performing and Recording
- ✓ Analyzing and Interpreting

Materials

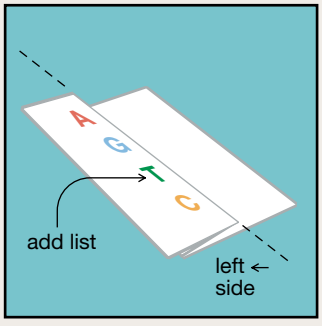
- blank piece of paper
- pencil

Procedure

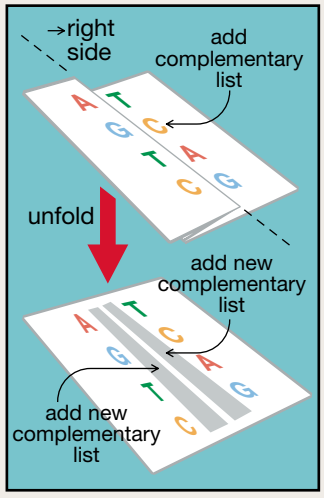
step 1: Fold the piece of paper as shown on the following illustration.



step 2: On the left side of the fold, record a random list of 15 nitrogen bases. Use the initials A, C, T, and G to represent each base.



step 3: On the right side of the fold, record the complementary strand of base pairs to complete the DNA molecule.



step 4: Unfold the piece of paper and lay it out flat in front of you so that the two strands of bases are now separated. Add the complementary strand of base pairs to each of the separated strands.

step 5: Save the piece of paper to help you answer the "Analysis" questions.

Analysis

1. You now have three complete sets of DNA to compare. These are the original strand that can be viewed by re-folding the paper, and the two duplicate strands that can be seen by unfolding the paper and pressing it flat. Compare all three strands of DNA.
2. How do you account for the trends you identified in your answer to question 1?

DNA Replication

When a cell needs to multiply to aid in the growth or repair of an organism, its DNA must first be copied. It is important that each of the two daughter cells produced during mitosis receives an exact copy of the parent cell's DNA. The process of making an extra copy of DNA for a new cell is called DNA **replication**. This is illustrated in Figure A2.14.

During replication, the two complementary strands of DNA are untwisted, the bonds between the bases are broken, and the strands separate like a zipper being unzipped. This process is controlled and aided by several protein enzymes.

replication: the process of making two DNA molecules from one original molecule prior to cell division

The bases of free-floating nucleotides in the nucleus attach to the complementary bases of nucleotides exposed along the halves of the unzipped DNA. This process creates two new DNA molecules, each with one strand of the original DNA molecule and one newly created strand.

The newly paired strands coil back into the double-helix shape as they are formed. Working from each one of the separated original DNA strands to make two new molecules makes DNA replication less prone to errors.

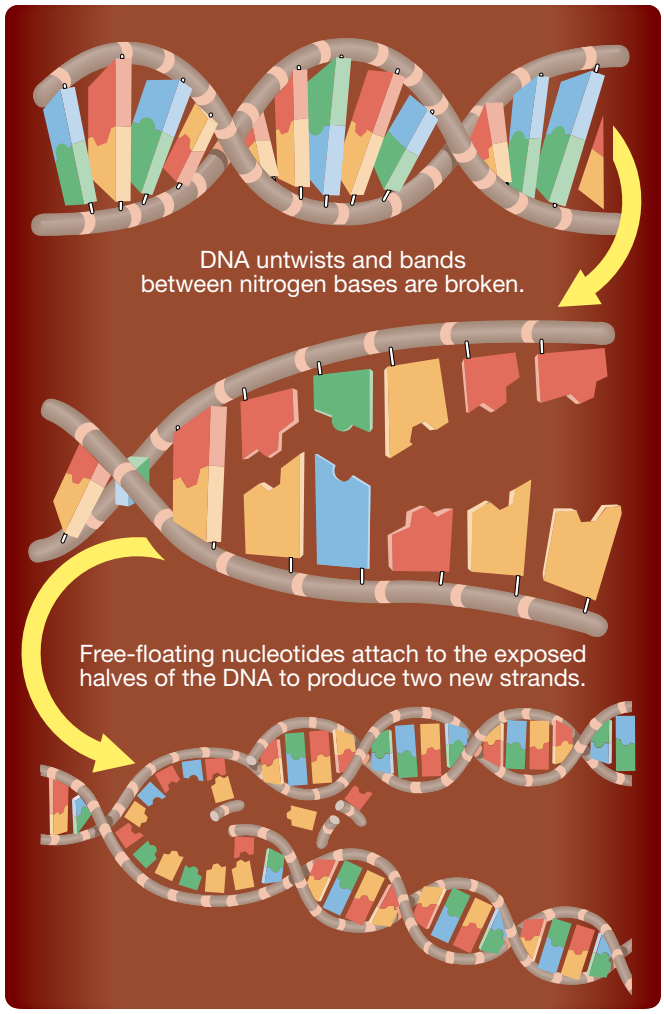
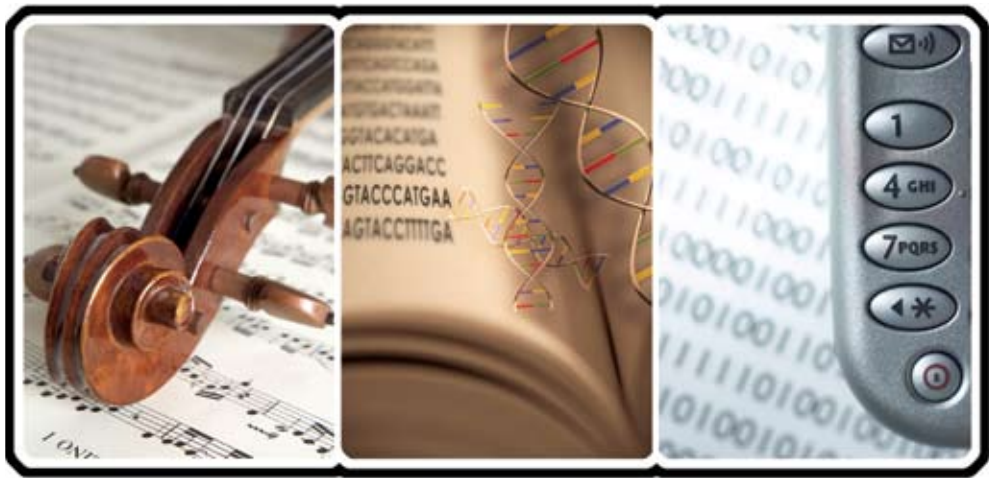


Figure A2.14

The Genetic Code



You may have been surprised to discover that only four different nucleotides make up DNA. How can such complex instructions—like the gene to make a protein that helps to clot blood—be written by so few letters?

Think of how the many English words you read every day are all made up of different combinations of just 26 letters. The order in which these letters are put together, combined with the spaces and punctuation between them, can form words, sentences, and instructions meaningful to the reader. Some forms of communication, such as musical notes, Morse code, or the binary language of computers, can be accomplished by using far fewer than 26 letters. Morse code, which is a series of dots and dashes, was used to pass messages along telegraph lines and until recently was used for visual communication between ships.

Binary language is made up of just two digits: 0 and 1. By using these two digits, a computer can be told how to carry out complicated operations. DNA code works on the same principle as other code systems. The arrangement of nucleotides on the DNA molecule creates a meaningful information sequence able to direct complicated processes in your cells.

Using the Genetic Code—Protein Synthesis

amino acid: one of the 20 possible building blocks of proteins determined by the genetic code of DNA

DNA triplet code: three adjacent nitrogen bases found on a gene that codes for the amino acid to be produced, begin, or end the reading of a gene

DNA, sheet music, and cookbooks are all sets of instructions. The DNA molecule does not actually make proteins, any more than a sheet of music plays a song or a cookbook makes a cake. Products can be made by following the step-by-step instructions of a recipe, a sheet of music, or a gene.

In Lesson 2.1 you learned that genes are regions along a DNA molecule. A gene is like a recipe for making a protein. A person doesn't have to read through each recipe in a cookbook to make a desired recipe. Similarly, the entire DNA doesn't have to be read to make one protein, but only the part where the gene occurs.

Proteins are chain-like molecules composed of smaller units called **amino acids**. Amino acids are like the individual links in the protein chain or the ingredients that get put together to make a recipe. The instructions contained on the gene are written in three-letter groups, with each group acting like a word. Any combination of three of the four DNA nitrogen bases (adenine, cytosine, thymine, or guanine) forms a **DNA triplet code**. Each group of triplet bases stands for an amino acid. For example, the DNA triplet code AAA corresponds to the production of the lysine amino acid. The amino acid produced for each triplet is summarized in "DNA Triplet Codes and Their Corresponding Amino Acids."

Even though there are 64 possible combinations of three DNA bases that can be composed from the four bases found in DNA, only 20 amino acids are used to construct amino acid chains in humans. This allows for some code redundancy. For example, the DNA triplet code AAG and the DNA triplet code AAA both correspond to lysine amino acid. These two DNA triplet codes are like synonyms. One of the DNA triplet codes, ATG, acts like the capital letter at the beginning of a sentence. The ATG triplet code marks the place where a gene is to start being read. There are also three DNA triplet codes, known as TAA, TAG, and TGA, which act like periods at the end of a sentence. They mark the place where the gene finishes being read.

DNA Nitrogen Bases

Nitrogen Base	Abbreviation
Adenine	A
Cytosine	C
Guanine	G
Thymine	T

DNA Triplet Codes and Their Corresponding Amino Acids

FIRST BASE	SECOND BASE: T		SECOND BASE: C		SECOND BASE: A		SECOND BASE: G		THIRD BASE
T	TTT	Phenylanine	TCT	Serine	TAT	Tyrosine	TGT	Cysteine	T
	TTC	Phenylanine	TCC	Serine	TAC	Tyrosine	TGC	Cysteine	C
	TTA	Leucine	TCA	Serine	TAA	STOP**	TGA	STOP**	A
	TTG	Leucine	TCG	Serine	TAG	STOP**	TGG	Tryptophan	G
C	CTT	Leucine	CCT	Proline	CAT	Histidine	CGT	Arginine	T
	CTC	Leucine	CCC	Proline	CAC	Histidine	CGC	Arginine	C
	CTA	Leucine	CCA	Proline	CAA	Glutamine	CGA	Arginine	A
	CTG	Leucine	CCG	Proline	CAG	Glutamine	CGG	Arginine	G
A	ATT	Isoleucine	ACT	Threonine	AAT	Asparagine	AGT	Serine	T
	ATC	Isoleucine	ACC	Threonine	AAC	Asparagine	AGC	Serine	C
	ATA	Isoleucine	ACA	Threonine	AAA	Lysine	AGA	Arginine	A
	ATG	MET or START*	ACG	Threonine	AAG	Lysine	AGG	Arginine	G
G	GTT	Valine	GCT	Alanine	GAT	Asparate	GGT	Glycine	T
	GTC	Valine	GCC	Alanine	GAC	Asparate	GGC	Glycine	C
	GTA	Valine	GCA	Alanine	GAA	Glutamate	GGA	Glycine	A
	GTG	Valine	GCG	Alanine	GAG	Glutamate	GGG	Glycine	G

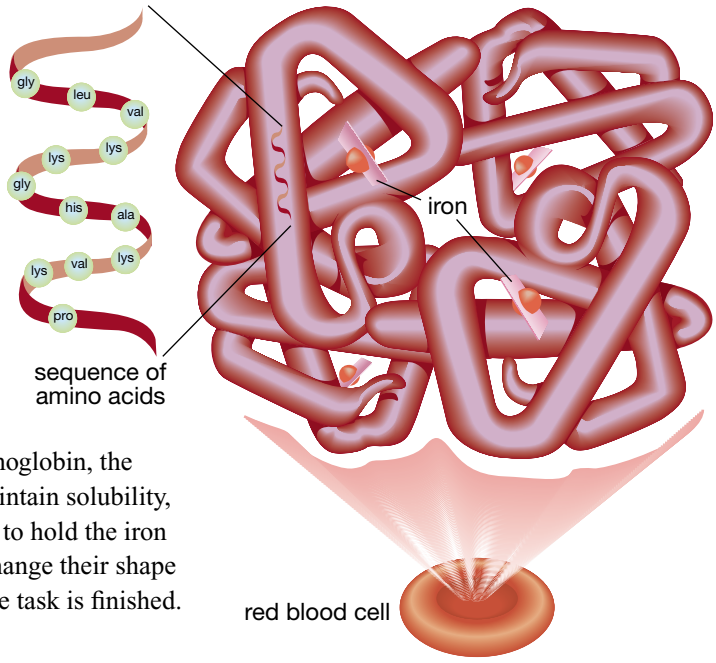
* ATG is an initiator triplet code and also codes for the amino acid methionine.
** TAA, TAG, and TGA are terminator triplet codes.

The Versatility of Proteins

Earlier in this unit you learned that proteins are molecules that serve a variety of useful and important bodily functions. Hemoglobin is the oxygen-carrying protein in red blood cells. Like every other protein, hemoglobin is a molecule composed of one or more chains of amino acids. The unique sequence of amino acids within this protein, combined with its particular coiled shape, accounts for the protein's distinct properties.

Proteins have one or more amino acid chains and are coiled into a variety of shapes. The versatility of an amino acid chain means that protein structure can take on a variety of forms to suit different body needs. The way that the chains of amino acids are folded and combined together determines the protein function. In the case of hemoglobin, the amino acids on the outside of the hemoglobin molecule maintain solubility, while special amino acids on the inside of the molecule act to hold the iron compounds that bind to oxygen. Some proteins can even change their shape to perform a task and return to their original shape when the task is finished.

Hemoglobin—A Protein Molecule



Practice

33. List the amino acid sequence that would be produced from the following base sequence found on a gene segment.

- **a.** ATAAAGCGACTTCCC
- **b.** AGAGGGGGTCTAGCC
- **c.** GTATTAGATTACGTTACA

34. Write a DNA sequence of bases that coded for the production of the following amino acid chains.

- **a.** Tryptophan-Phenylanine-Tyrosine
- **b.** Methionine-Glutamate-Aspartate
- **c.** Glutamate-Methionine-Cysteine

Utilizing Technology

Interpreting the Genetic Code

Science Skills

✓ Performing and Recording

Purpose

To construct a sequence of amino acids, you will decipher the coding along a segment of DNA.

Procedure

Locate the applet called "DNA—The Genetic Material," on the Science 30 Textbook CD. Follow the instructions.

2.3 Summary

Deoxyribonucleic acid, or DNA, is the molecule that contains the coded instructions for creating proteins. Genes are regions along the DNA that code for a specific protein. A nucleotide is a chemical unit made up of a phosphate, a deoxyribose sugar, and a nitrogen base.

There are four nitrogen bases: adenine, thymine, cytosine, and guanine. Adenine only bonds to thymine and cytosine only bonds to guanine (A-T or T-A and C-G or G-C). When one base is bonded with its complementary base, the two bases are called a base pair.

DNA structure is two strands of nucleotides attached by their complementary bases and twisted into a double-helix shape. DNA has to be wound very tightly to fit into the nucleus. The molecule must become very twisted and it gets wound around proteins—called histones—that act like spools. To replicate, DNA pulls apart and complementary free-floating nucleotides attach to the appropriate exposed bases of the strands to create two new molecules, each with half of the original DNA molecule.

Within a gene, the nitrogen bases are read as triplets. Each triplet provides information about the formation of a polypeptide chain of amino acids used to make proteins.

2.3 Questions

Knowledge

1. Indicate whether each of the following statements is true or false. If a statement is false, explain why.

a. A DNA triplet code is made up of three amino acids.

b. A DNA triplet code may code for the same amino acid as another DNA triplet code.

c. Adenine bases can only bond to cytosine bases.

d. A double helix is similar in shape to a spiral staircase.

e. Genes provide the instructions to make proteins.

f. There are ten different amino acids.

g. Histone is one of the four base pairs found along the DNA molecule.

h. Alternating phosphate and deoxyribose sugar make up the backbone of a DNA strand with the base pairs attached in the middle.

i. During replication, the DNA breaks into small pieces and re-forms as two smaller halves.

j. A DNA molecule has three strands of nucleotides braided together.

Applying Concepts

2. Use the "DNA Triplet Codes and Their Corresponding Amino Acids" table to determine which of the following DNA sequences would code for the production of valine-alanine-asparagine.

I. AAAAGAATA

II. CATCGCACA

III. GTGGCTAAT

3. Draw a series of diagrams to show how DNA is replicated. Use two colours to distinguish between the original DNA strands and the newly produced DNA strands.

4. Like every other protein, hemoglobin consists of chains of amino acids. The sequence of amino acids in one section of a hemoglobin molecule is Glycine-Leucine-Valine . . . . Determine the corresponding DNA triplet codes that would provide the instructions for building the first three amino acids in this sequence.

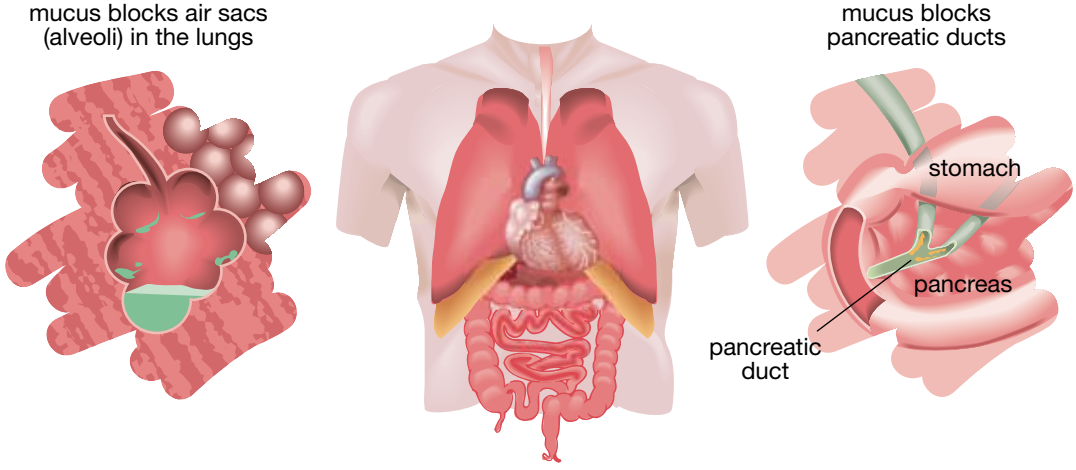
5. Complete the following table that compares protein synthesis to making a cake from a recipe.

Making a Cake	Protein Synthesis
• a library of cookbooks	• a karyotype of all the chromosomes for one individual
• a cookbook of recipes	
• a recipe for a particular cake	
• the words of the recipe	
• ingredients that go into the cake	
• the finished cake product	

2.4 Mutations and Genetic Diseases

Rain or shine, people will show their support for a fundraising walkathon. The Great Strides Walk is held annually to support the efforts of the Cystic Fibrosis Foundation. In 2006 this event raised more than $775 000, as thousands of Canadians participated in more than 40 locations.

The Effects of Cystic Fibrosis



Cystic fibrosis is an inherited condition. It affects the cells that produce the juices of mucus, sweat, saliva, and digestion. One of the functions of these secretions is to act as lubricants, so they normally have thin and slippery consistencies. In cystic fibrosis, a defective gene causes these secretions to become thick and sticky. The result is that they can plug up lung and pancreas passageways.

Earlier in Chapter 2 you learned that genes are like recipes or sets of instructions. You also learned that the sequence of bases in the DNA molecule is comparable to the letters in the words of a written recipe. Imagine that you are copying down a set of instructions in a recipe and you don't notice that you made a mistake. How could the mistake affect your finished product? What if a mistake is made during DNA replication and the base sequence of DNA is copied incorrectly? What effect might such a change have in the DNA sequence, its structure, or how it affects the protein product derived from this DNA molecule?

A change in the sequence of bases along DNA is called a **mutation**. When you hear the word *mutation*, you might think of a science fiction story or a horror movie where a dramatic event causes a character to become described as a mutant with a changed appearance or abilities. Most mutations are actually small changes to the DNA sequence that occur naturally, or are due to exposure to high energy radiation or chemicals. An important characteristic of mutations is that the changes to DNA are carried forward in subsequent DNA replications and are inherited by future generations.

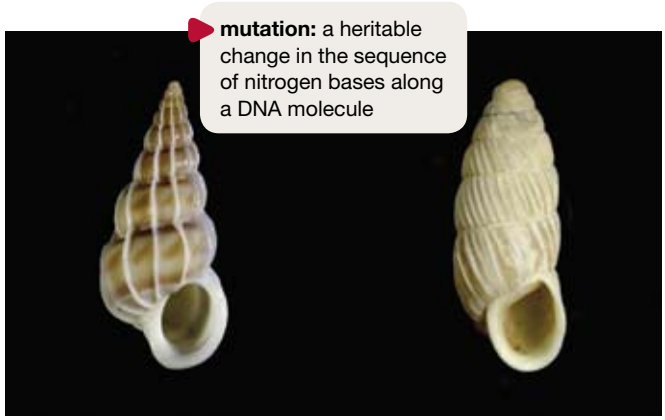


Figure A2.15: Mutations are responsible for variations within a species.

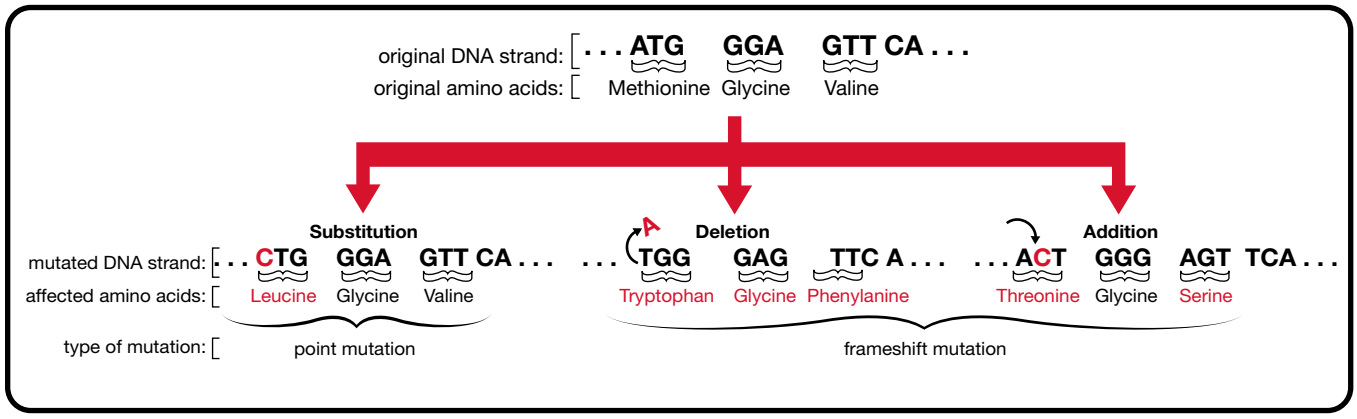
As you learned in previous science courses, mutations are the source of variation within wildlife populations. This variation leads to adaptations, which are acted upon by natural selection to drive evolutionary change. Mutations clearly play an enormous role in the study of biological systems.

In this lesson you will learn how mutations in DNA affect the proteins produced. This occasionally results in human diseases that can be inherited by offspring. Using Mendel's theories and Punnett squares, you will predict the probability of offspring inheriting a genetic disease. You will also learn how to read a pedigree chart that traces genetic diseases through families. Mutations that benefit an organism and mutations that result in resistance to bacteria will also be examined in this lesson.

Mechanisms of Mutation

Mutations are like "typos" in a word-processing document. For both recipes and genes, the effect that a copying mistake has on the product depends on where it happened and on what kind of mistake was made. For example, if you made a slight spelling mistake to the word *salt* and instead wrote *saltt* when copying a recipe, it wouldn't change how you carry out the recipe. And it would have little effect on the finished product. The recipe would still work. But if you wrote *25 mL of salt* instead of the original *2.5 mL of salt*, your recipe would most likely be ruined. In most cases, the mutation of DNA has little effect on the products produced from a gene—the mutations occur in chromosome regions that do not include genetic instructions, or the affected gene sequence is still able to function despite the mistake. Cells can usually repair minor DNA mutations that frequently occur in the human body. Mutations to DNA can occasionally cause the gene to stop working or to work differently, as is the case with cystic fibrosis.

How Mutations Affect Genetic Information



Mutations in DNA can happen when one nucleotide—during replication—accidentally gets substituted for another nucleotide. The chart "How Mutations Affect Genetic Information" illustrates what happens when there is a substitution of one nucleotide—ATGGGAGTT changes to become CTGGGAGTT. The amino acid chain produced from this sequence is now altered, possibly affecting the protein functioning. A nucleotide base substitution is called a **point mutation**, which is similar to a typo where one letter of a word gets changed and often alters the word's meaning. If the phrase "the fox can run" was mistyped as "the box can run," the single letter substitution not only changes the meaning of the phrase but it makes the phrase confusing.

The deletion or addition of a nucleotide can also affect the DNA base sequence resulting in an altered or incomplete amino acid chain. The results tend to be more serious in this case because all of the nucleotides "downstream" of the mutation are affected. In other words, the grouping into sets of three or the framing of all the nucleotides that follow is changed. That is why this is called a **frameshift mutation**. If the phrase "the fox can run" had an extra letter added at the beginning of the phrase but the phrase was still separated into three-letter words, it would read "ath efo xca nru n." The frameshift mutation turns the original phrase into nonsense.

point mutation: the substitution of one nucleotide base for another during DNA replication

frameshift mutation: the deletion or addition of a nucleotide during DNA replication
This change causes the three-letter groupings or frames in DNA to be read in an alternate pattern.

Practice

35. Figure A2.16 shows two DNA strands—the lower strand is a product of replication. A mutation occurs in the middle of a gene sequence. The sequence of bases gets changed from TAT to TAA.

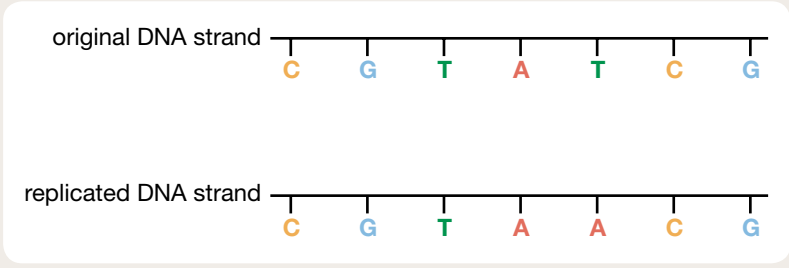


Figure A2.16

- **a.** Is this a point mutation or a frameshift mutation?
- **b.** Use your table "DNA Triplet Codes and Their Corresponding Amino Acids" to identify what amino acid corresponds with the DNA sequence of TAT.
- **c.** What corresponds with the DNA sequence of TAA?
- **d.** Explain what effect the change to the DNA sequence has on the production of the amino acid chain.

36. A gene sequence reads GGATTAGAG. A mutation occurs and the sequence now appears as GGGATTAGAG.

- **a.** Identify the sequence change as either a point mutation or a frameshift mutation.
- **b.** Use your table of DNA triplet codes to list the amino acid sequence produced by the original DNA strand.
- **c.** Use your table of DNA triplet codes to list the amino acid sequence produced by the new, mutated strand.
- **d.** Explain the effect of this sequence change on the production of the amino acid chain.

Passing On Mutations

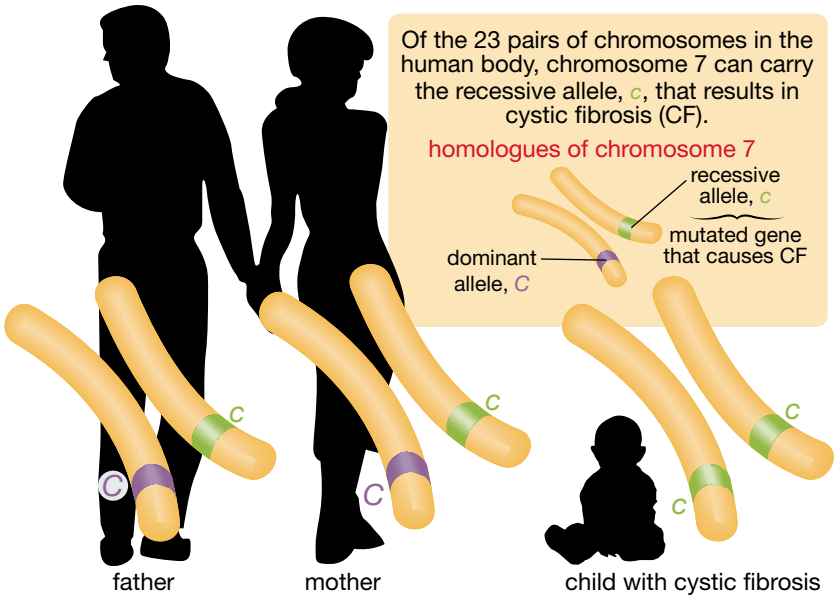
When you think of a disease you probably think of an illness spread by a disease-causing pathogen, but some illnesses result from the presence of one or several mutated genes. Initially, a mutation can be caused in one individual by an exposure to something in the environment, such as X-rays, ultraviolet radiation, toxic chemicals, or some other factor that causes a change to the nitrogen bases in DNA. The mutations that result often have little effect on the functioning of our bodies. However, in other instances the mutation impairs the function of a gene or the amino acid chain produced, which results in negative consequences for the individual.

If a mutated gene is present in one body cell and that cell is no longer able to fulfill its role, the cell usually dies and other body cells of the same type compensate for the loss of one faulty cell. Recall that there are not only body (somatic) cells but sex cells, such as eggs and sperm. What effect could a mutation have in the genes of a sex cell?

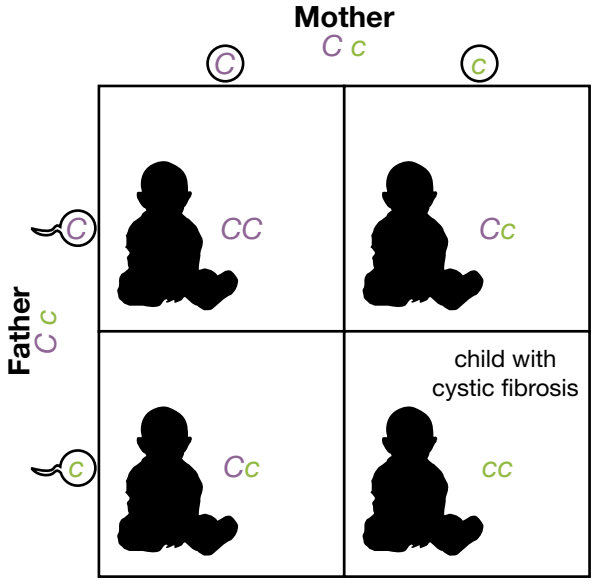
If a mutation is capable of being passed between generations and if that mutation results in illness, then the resulting condition is called a **genetic disease**. Cystic fibrosis is a genetic disease passed on from parents to their children. In this case, chromosome 7 carries the mutated gene that results in the disease.

When the gamete with the mutation combines with the gamete of the other parent to produce offspring, the mutated gene is present in every cell of the newly developing offspring. The presence of this mutated gene in each cell can cause the offspring to develop abnormally, die at an early age, or develop a genetic disease. Recent advances in medications and therapies can allow an individual to live longer with an improved quality of life. In spite of this, genetic diseases like cystic fibrosis are difficult to treat and cure because the illness is caused by a mistake in the genes of every body cell, rather than a foreign invading pathogen.

Having one set of genes from each parent can result in a decreased incidence of genetic disease. In many genetic diseases, symptoms are associated with a mutation. The inheritance of a non-mutated copy of a gene from one parent can often compensate for the mutations present in the gene inherited from the other parent. This is why some individuals carry a mutated gene but do not develop the symptoms of a genetic disease. The non-mutated copy of the gene that they received from their other parent is working properly to produce the necessary protein to keep them healthy. An individual who possesses a disease-causing, mutated copy of a gene but who does not develop the symptoms of that genetic disease is called a **carrier**. When a carrier of a genetic disease has a child, there is an increased chance of the child developing the genetic disease. When doctors and other medical practitioners ask about your family history of certain diseases, they are trying to determine the probability of you developing genetically inherited diseases or conditions.



Each parent carries the dominant allele, *C*, and the recessive allele, *c*. A Punnett square can be used to show that the probability of their offspring inheriting two recessive alleles is ¼ or 25%.



Note that both parents and probably half of the children will be carriers of cystic fibrosis because they have the genotype *Cc*.

genetic disease: a disease caused by a mutation of one or more genes that can be inherited by future generations

carrier: an individual who possesses a form of a gene (allele) that results in a disease but does not demonstrate, exhibit, show, or have the symptoms of that disease

However, this individual can pass the disease-causing allele to his or her offspring.

Practice

37. In your health file, indicate any genetic diseases that run in your family. Some examples of genetic diseases include the following: Tay-Sachs disease, sickle cell anemia, phenylketonuria (PKU), Huntington disease, hemophilia, cystic fibrosis (CF), albinism, Marfan syndrome, polycystic kidney disease, Zellweger syndrome, Adrenoleukodystrophy (ALD), achondroplasia, and maple syrup urine disease.

If you are not sure whether a medical condition in your family is a genetic disease, you can try entering the phrase, "Is _____ a genetic disease?" in your Internet search engine.

Patterns of Genetic Disease Inheritance

Genetic diseases can be caused by alleles that behave similarly to other recessive or dominant alleles, and they can be autosomal or sex-linked in their mechanisms of inheritance. If the genetic disease is caused by a recessive allele, what genotype would a person with the genetic disease possess? A dominant allele?

If the disease is caused by a recessive allele, an individual requires two copies of a mutated recessive allele (homozygous recessive) to develop the disease. If caused by a dominant allele, only one copy of the mutated allele (heterozygous dominant or homozygous dominant) is needed to cause the disease to develop. Check the following table.

TABLE OF GENETIC DISEASES

Genetic Disease	Symptoms	Location of Gene	Mechanism of Inheritance	Prevalence
cystic fibrosis (CF)	People with CF produce thick sticky mucus that builds up in their lungs and digestive tract. This makes it difficult to properly breathe and digest food. People with CF are also prone to lung infections because they cannot easily clear bacteria from their lungs.	chromosome 7	autosomal recessive	Approximately 1 in 2500 children born in Canada has CF and 1 in 25 Canadians is a carrier of the defective allele that causes cystic fibrosis.
Huntington disease (once called Huntington's chorea)	Huntington disease causes brain cells to die in particular regions. This results in a continual reduction in the ability to control movements, remember events, make decisions, and control emotions. Symptoms usually appear between the ages of 30 and 45.	chromosome 4	autosomal dominant	Approximately 1 in 10 000 Canadians has Huntington disease.
hemophilia	There are two forms of this disease: hemophilia A and hemophilia B. Both forms are caused by a mutation of one of the genes that produces blood-clotting proteins. Both of the genes involved with producing the proteins for blood clotting are found on the *X* chromosome. A defective allele for either of these two *X*-chromosome genes can result in impaired blood-clotting ability. People with hemophilia bleed for a longer time period than people without this condition. Internal bleeding, or hemorrhaging, is a common risk associated with this dangerous condition.	*X* chromosome	sex-linked recessive	Hemophilia A affects about 1 in 10 000 people in Canada and hemophilia B affects as few as 1 in 50 000 people. Because of the sex-linked nature of the disease, males develop the disease more than females do.

Practice

Use the following information to answer questions 38 to 40. When genetic diseases are controlled by a single pair of alleles, the patterns of inheritance described by Mendel's studies and depicted using Punnett squares can help determine the probability of offspring developing a genetic disease. For each of the following crosses involving the inheritance of genetic diseases, choose letters to represent alleles, draw a Punnett square for each cross, and answer the questions about the cross.

38. A couple discover that they both have a family history of cystic fibrosis. They are thinking of having a child, and they ask for a genetic test to be done. Both the man and the woman discover that they are carriers of the recessive cystic fibrosis allele.

- **a.** Build a Punnett square to describe this cross.
- **b.** What is the percentage probability that their child will develop cystic fibrosis?
- **c.** What is the percentage probability that their child will be a carrier of the cystic fibrosis allele?
- **d.** What is the percentage probability that their child will not inherit the cystic fibrosis allele?

39. A man is heterozygous for the dominant Huntington allele, and he has a child with a woman who does not have a Huntington allele.

- **a.** Build a Punnett square to describe this cross.
- **b.** Write the possible offspring genotypes from this cross.
- **c.** What is the percentage probability that their child will not develop Huntington disease?
- **d.** What is the percentage probability that their child will develop Huntington disease?

40. A woman carries one of the defective recessive alleles on her *X* chromosome that causes hemophilia. She has a child with a man who does not possess the hemophilia allele.

- **a.** Build a Punnett square to describe this cross.
- **b.** What is the percentage probability that she will have a child with hemophilia?
- **c.** What percentage of females born from this cross are likely to have hemophilia?
- **d.** What percentage of males born from this cross are likely to have hemophilia?

41. Use the Internet to gather information about the specific organizations in Canada that raise funds and provide support for persons with the following genetic diseases.

- **a.** cystic fibrosis
- **b.** Huntington disease
- **c.** hemophilia

In each case, find the organization's website. Use the site to determine the organization's mission statement and opportunities for people to volunteer or make donations.

Factors That Increase Mutations

Environmental influences can affect the frequency with which mutations occur naturally. People are exposed to some chemicals and electromagnetic radiation that have been shown to increase the frequency of mutations that occur in the human body. An environmental influence that increases the chance of mutation is called a **mutagen**. A mutation in body cells may cause a mistake in the genes that control cell division, resulting in the uncontrolled division of body cells. The abnormal and uncontrolled division of body cells is called *cancer*, and a mutagen known to cause cancer is called a **carcinogen**. Common carcinogens include many chemical agents found in ionizing electromagnetic radiation—such as ultraviolet rays and X-rays —as well as agents in cigarette smoke and pesticides.

- **mutagen:** any agent that causes the likelihood of mutations to increase
- **carcinogen:** any agent that causes the likelihood of cancer to increase

Many carcinogens are also mutagens.

Science Links

What's the definition of a cigarette? One official in the health-care field described a cigarette as ". . . a delivery system for toxic chemicals and carcinogens."

There are more than 4000 chemicals found in cigarette smoke. And 40 or more of them are known carcinogens, including benzene—a petroleum solvent—and formaldehyde, which is used to preserve dead bodies. In Unit B you'll learn more about the chemistry of some of these toxic organic compounds.

Tracing Genetic Disease: Pedigree Charts

If you observe an individual with a dominant trait, such as the ability to roll her tongue, can you tell if that individual is homozygous or heterozygous for the dominant allele? Is it possible to tell if this person carries a recessive allele? Is there a simple way to detect whether his or her genotype is *Rr* or *RR*? Unfortunately, there is no way to do so by only looking at this specific person, but if you can look at the individual's family, sometimes the answer is staring directly at you. For example, if two tongue-rolling individuals have a child without tongue-rolling ability, it indicates that they both carry the recessive gene for tongue rolling. They would both have to be heterozygous ($Rr \times Rr$) to produce a child who is homozygous recessive (rr).

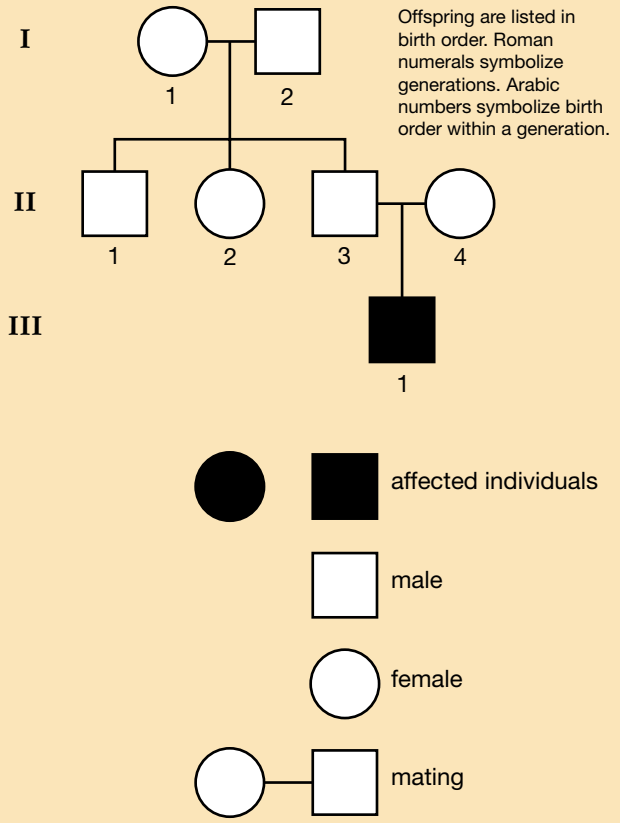
Geneticists use a tool called a **pedigree** to predict the genotype of an individual. A pedigree is like a genetic family tree. You may have previously heard the term in relation to dog breeding or horse breeding. This is because many animal breeders keep detailed lineage records of the animals they breed, and then they use the pedigrees to trace specific traits. A pedigree is a useful technology for tracing genetic diseases.

Genetic pedigrees use a specific set of symbols to identify known genotypes of family members so that unknown genotypes can be predicted. Circles are used to represent females, and squares are used to represent males. On some pedigree charts, a shaded individual indicates a person with the condition being studied. Individuals who are known carriers are sometimes identified by being drawn as half-shaded. A line drawn between two individuals indicates that they have had offspring. Roman numerals and a new row are used to indicate each generation, and individuals are numbered within each generation.

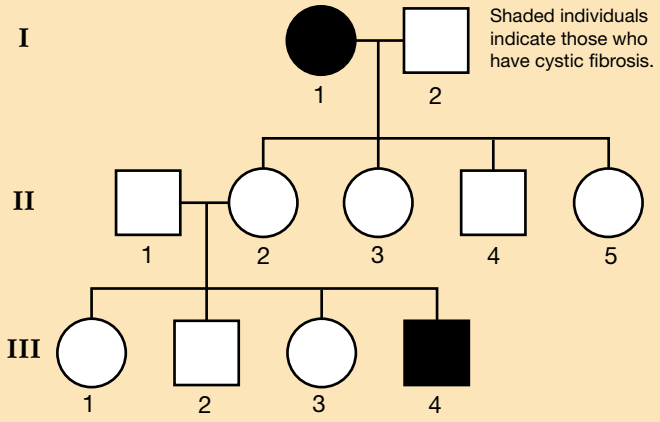
The "Cystic Fibrosis Pedigree" is for a family with members who have the autosomal recessive genetic disease known as cystic fibrosis.

The "Cystic Fibrosis Pedigree" reveals that a first-generation couple had four children—three girls and one boy. The shading indicates that the first generation female (I-1), or the grandmother, had cystic fibrosis. This means that she had to possess two recessive alleles for the condition. The pedigree also shows that none of her four children developed the disease, since the symbols to represent them are not shaded. Because the mother only has the recessive allele to donate, her children all received the allele for cystic fibrosis. Each of her children is a carrier for the disease. Her eldest daughter (II-2) had four children, and the youngest son (III-4) of this daughter developed the disease. In order for this grandson to have the disease, his father (II-1) also has to be a carrier of the recessive allele.

A Guide to Pedigree Charts



Cystic Fibrosis Pedigree
(Autosomal Recessive)



pedigree: a set of standard symbols used as a tool for geneticists to trace a particular trait

It is like a genetic family tree.

The pedigree in Figure A2.17 is for a family with members having the autosomal dominant genetic disease called Huntington disease. A trait that re-appears in each successive generation, as with Huntington disease, is usually caused by a dominant allele.

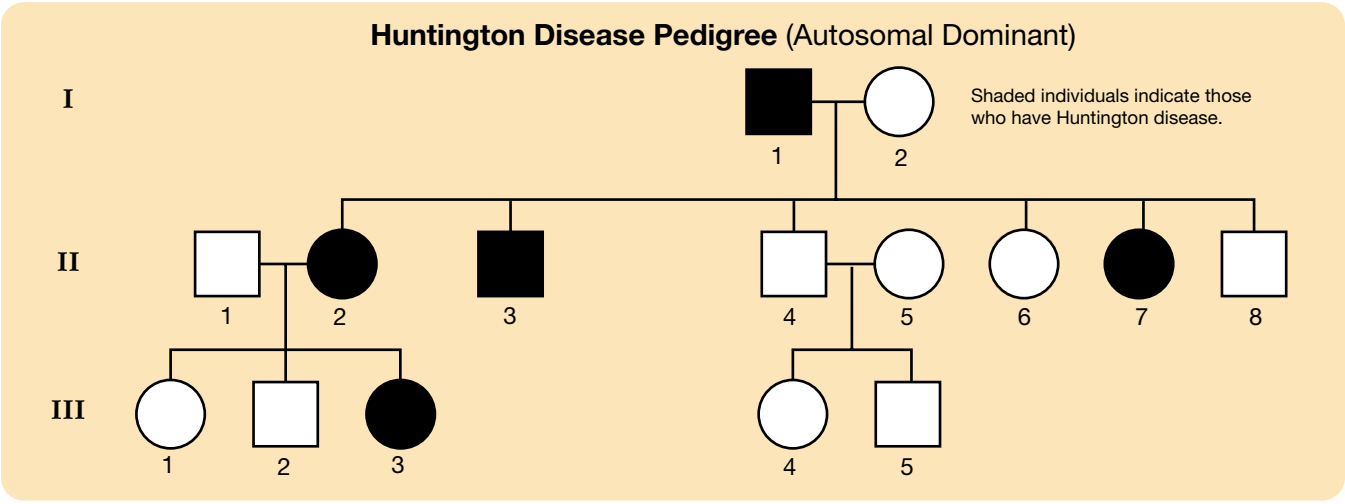
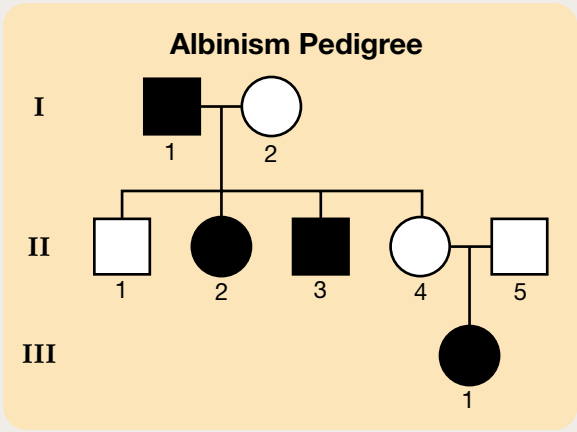


Figure A2.17

Practice

42. Albinism is a genetic condition that causes an absense of pigmentation in skin, hair, and eyes. In humans, the most severe form of albinism—called *oculocutaneous albinism*—is an autosomal recessive genetic disease. Examine the "Albinism Pedigree" and answer questions 42.a. to 42.c.



a. In the pedigree shown, determine the number of females with the albino condition. Determine the number of males with the albino condition.

b. Describe the phenotype of the individual (III-1) and her parents. Account for the differences between phenotypes in these two generations.

Figure A2.18: albino wallaby

c. List the most likely genotypes with respect to the albino trait for the eight people shown in this pedigree.

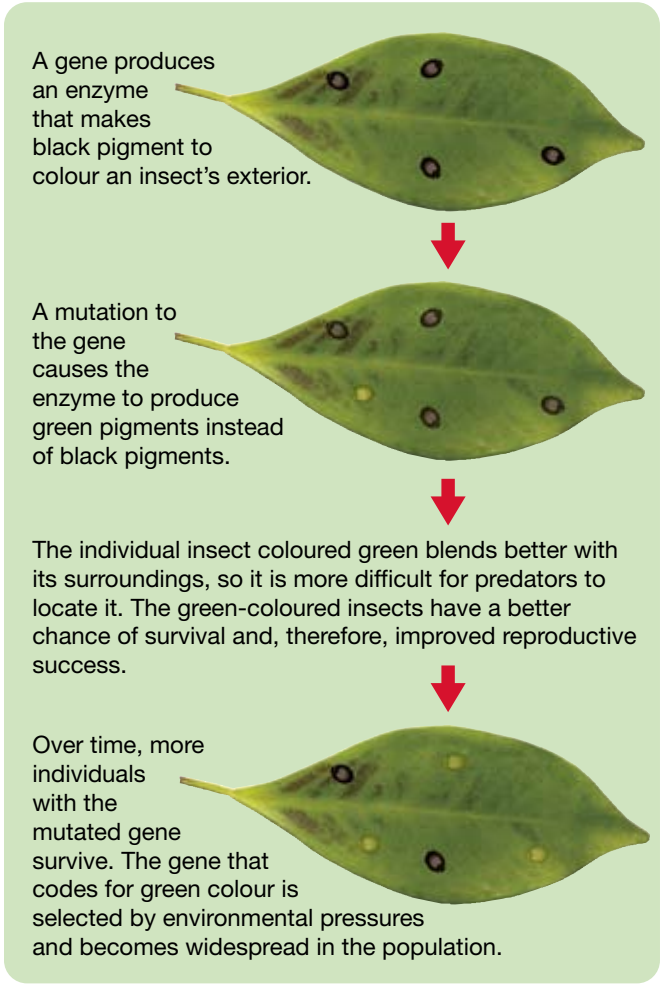
43. Sonja is not able to roll her tongue but her brother, Mikail, can roll his tongue. When Sonja surveys her family members, she finds that her mother is a tongue roller, and her father is not. Her father's only sister and brother cannot roll their tongues either. The two grandparents on Sonja's father's side are also non-tongue rollers. For the grandparents on the mother's side, the grandfather is a tongue roller, but the grandmother is not. Develop a pedigree for the family described.

44. Phenylketonuria (PKU) is a genetic disease caused by an inability to produce an enzyme. This missing enzyme causes a buildup of an amino acid in the body to toxic levels—this can result in organ damage and impaired intellectual development. Draw a pedigree for the following description of a family's genetic history for the autosomal recessive disease of phenylketonuria.

A male who does not exhibit PKU and a female who does not exhibit PKU have four boys. The two oldest sons are carriers of the recessive allele, and the youngest son develops the genetic disease. The third son neither has the disease nor is he a carrier. The oldest son has two daughters with another PKU carrier—both of these daughters develop the PKU genetic disease.

Beneficial Mutations Affect Populations—Evolution

You have learned that most mutations have little effect on an organism's functioning. In rare cases a mutation can impair a gene's ability to produce a protein, and this results in the disease symptoms. A mutation occasionally creates an advantage for the individual.



Perhaps the mutation causes a brighter colour of flower to be produced, which makes the flower with the mutation more successful at attracting pollinators.

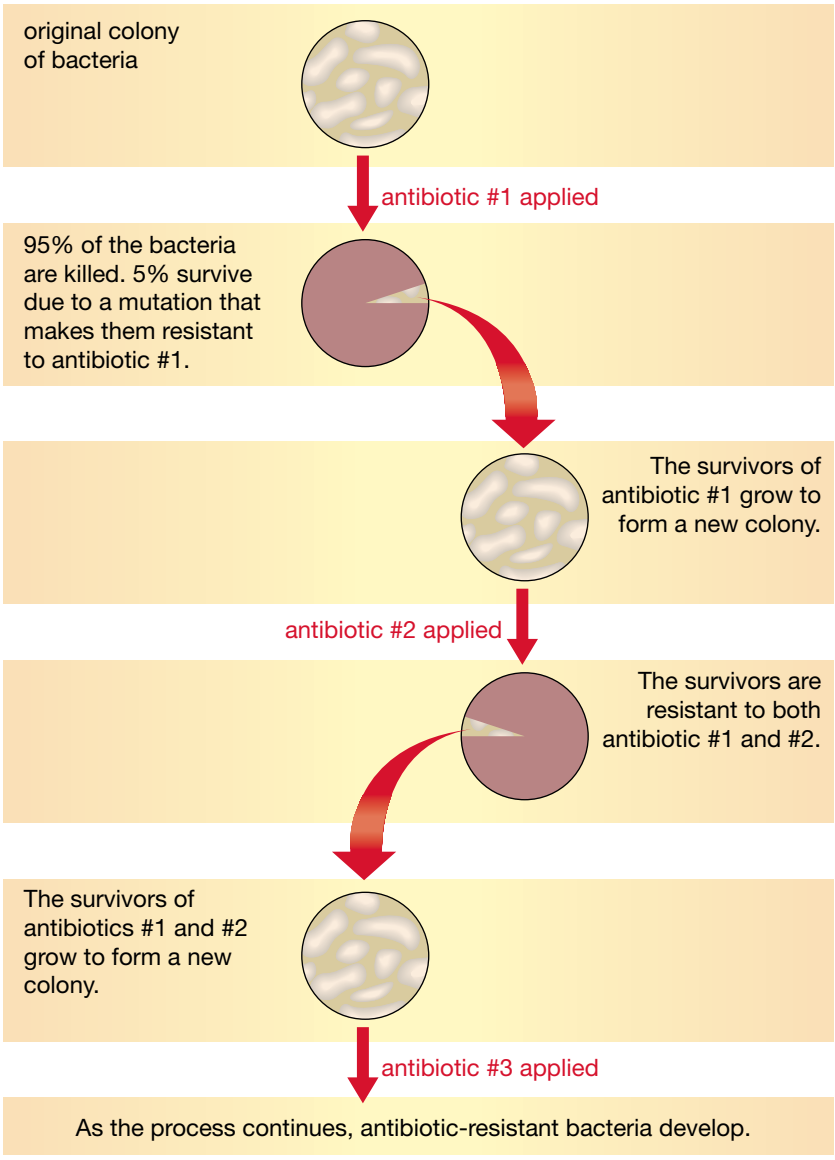
Or perhaps a mutation in genes controlling its colour provides the individual with improved camouflage to help protect it from predators.

In cases where mutations result in an advantage to the organism's survival, organisms possessing the mutation have a better chance to survive and breed. This means that this new variety is able to generate a greater number of offspring.

Perhaps you may recall from previous science courses that Charles Darwin used the term "survival of the fittest." This refers to the idea that the organisms best able to survive have the opportunity to reproduce and pass along their traits to their offspring. Mutations play a key role in this process of natural selection because they introduce the new alleles selected by the environment as being more favourable. In this way, advantageous traits become more widespread in a population and change a population's overall characteristics.

Resistance in Bacteria

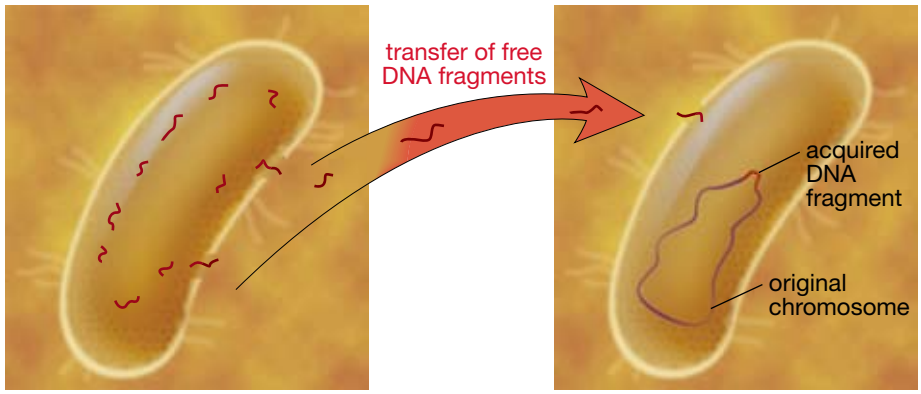
Antibiotics are chemicals that have saved countless lives by killling bacteria that cause infections and illnesses. When antibiotics are used, most of the bacteria die. Recently, the number of infectious bacteria resistant to antibiotics has increased. You have seen how mutations can result in the change or evolution in a species. Can mutations in bacteria be the cause of an increase in resistance to antibiotics?



If some bacteria possess a mutant gene that makes them resistant to antibiotics, they will not be killed. The resistant bacteria not killed by the antibiotic are the only individuals remaining to reproduce. Over time the new population—or strain—of bacteria consists almost entirely of members that possess genes resistant to that antibiotic. Each time a bacterial population is exposed to a new antibiotic, this process of natural selection repeats and only the resistant survive. Many scientists and doctors are becoming worried about the development of bacterial strains that are resistant to antibiotics. For this reason, antibiotics are prescribed less frequently than before, and patients who are prescribed antibiotics are advised to take all their medication even after they have begun to feel better to make certain a large percentage of the bacterial population is killed.

The process of antibiotic products "selecting" resistant bacteria is based not only upon the same principles of selective breeding that you studied earlier in this chapter, but upon the same principles of natural selection that you examined in earlier science courses. The main idea here is that individuals most suitable to a particular environment live to breed and pass on their genes.

Transformation of DNA Fragments

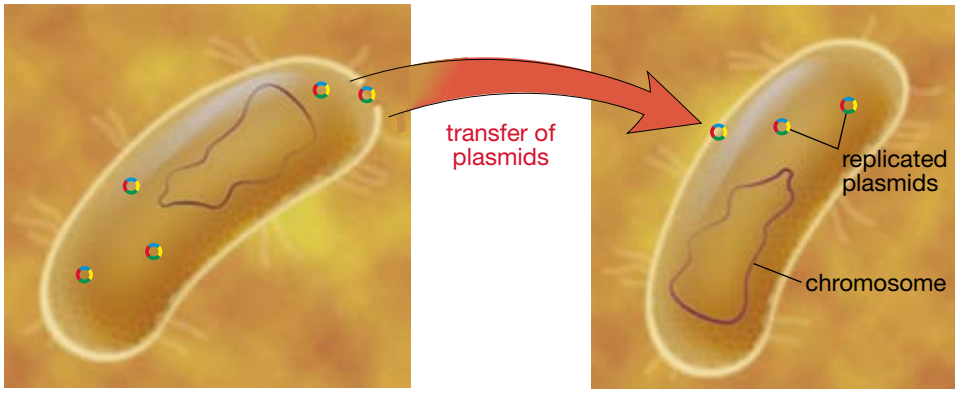


a dead bacterium

a living bacterium acquiring DNA from environment

As you learned earlier, evolution takes place over long periods of time and involves many generations. Because bacteria reproduce very quickly—as fast as once every 20 minutes—the speed of their evolution can appear to be rapid. The appearance of bacterial superbugs resistant to several kinds of antibiotics has made the scientific community question the exact mechanism for the evolution of this trait. Even though bacteria reproduce asexually, they have methods of exchanging DNA that create more genetic diversity and can result in the development of new traits, such as antibiotic resistance. Some bacteria can acquire foreign DNA from their surrounding environment and incorporate it into their own DNA in a process known as **transformation**.

Transformation of Plasmids



a living bacterium with plasmids

a bacterium acquiring plasmids

Many bacteria can also possess a small circular piece of DNA molecule, called a **plasmid**, which is separate from the DNA in the bacteria's chromosome. Plasmids are self-replicating and some have the ability to temporarily join to the bacterial DNA. The plasmids may only have a few genes not necessary for the regular functioning or survival of the bacteria, but these genes may influence other traits or lead to advantageous properties for the organism—this may include antibiotic resistance. Bacteria with a plasmid containing genes that provide antibiotic resistance make the bacteria exposed to antibiotics resistant to the drugs. Because plasmids can be transferred quite easily between bacterial cells when they contact one another, a plasmid can be transferred between individuals. This results in the sharing of antibiotic-resistant genes.

transformation: the process by which free DNA is incorporated into a bacterial cell

plasmid: a self-replicating circular piece of DNA that can be transferred between bacteria
Plasmid transfer allows for the sharing of genes on the plasmids between bacteria.

2.4 Summary

Mutations are changes in the sequence of DNA that can be inherited by future generations. These changes spontaneously occur and do not usually affect an individual. Mutations occasionally impair gene functioning or result in beneficial advantages. They can occur when nucleotides are substituted by point mutation, while deletions or additions of nucleotides result in frameshift mutation. Mutations that improve an organism's success within a population lead to adaptations selected by the environment. Natural selection is what drives the process of evolution. Resistance to antibiotic drugs has occurred in bacteria as a result of mutation, transformation, plasmid transfer, and natural selection.

A genetic disease is an illness resulting from faulty or impaired genes that can be inherited by future generations. The presence of two alleles for each gene in the human genetic make-up helps prevent the development of many genetic diseases. Individuals can be carriers of alleles that cause genetic diseases.

The pattern of inheritance for genetic diseases is the same studied previously for autosomal, sex-linked, dominant, or recessive alleles. Punnett squares can be used to predict the probability of offspring inheriting a genetic disease when the disease is caused by a single gene. Pedigrees can be used to trace the inheritance of a genetic disease in a family.

2.4 Questions

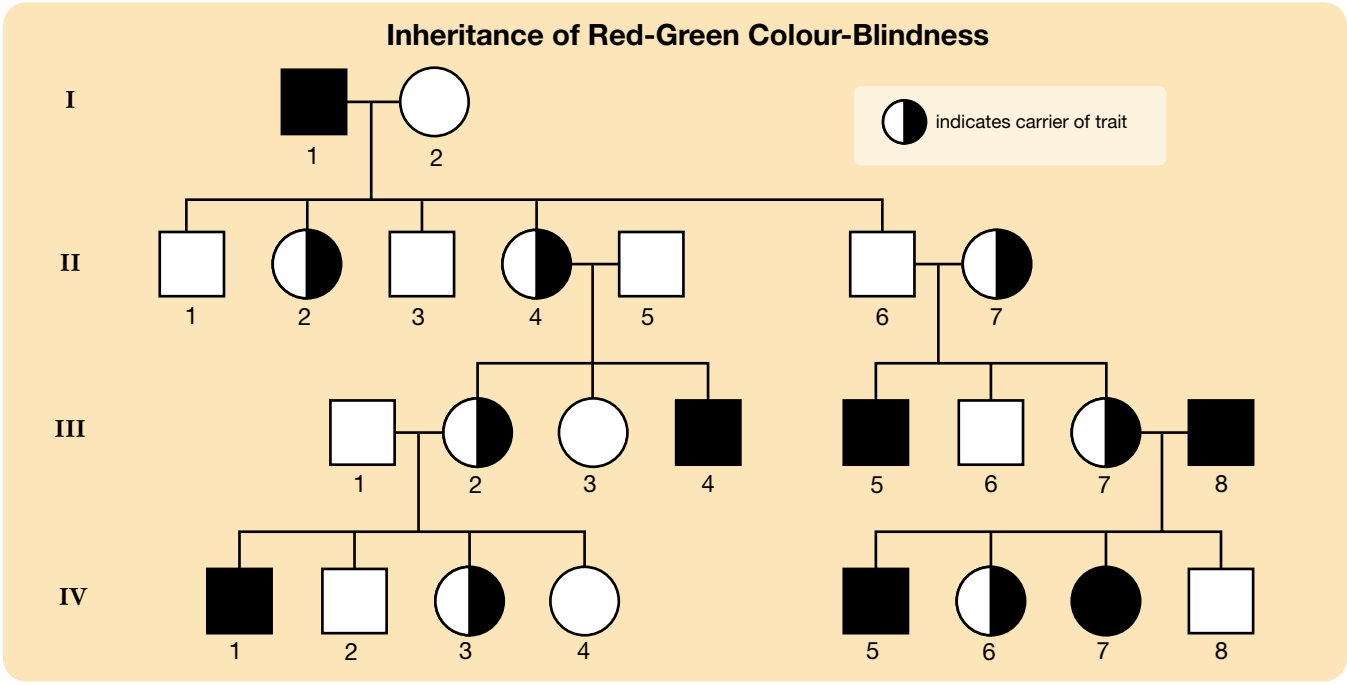
Knowledge

1. Define *mutation*.

2. Explain why a doctor may ask questions about the history of certain traits or illnesses in your family.

3. Explain how people who are carriers of the allele for cystic fibrosis do not have disease symptoms.

4. Red-green colour-blindness is a sex-linked trait. The pedigree in "Inheritance of Red-Green Colour-Blindness" shows the occurrence of the colour-blindness disorder for one family. Study the diagram and answer the following questions.



a. In the first generation, is the father or the mother colour-blind?

b. Determine the number of males and the number of females produced by the father and mother of the first generation.

c. State the number of individuals in this pedigree who are carriers for colour-blindness.

d. How many males and how many females have colour-blindness in this pedigree?

5. List several mutagens that can increase the frequency of mutations.

Applying Concepts

6. List two similarities and two differences between Punnett squares and pedigree charts.
7. Despite new therapies and other medical breakthroughs, cystic fibrosis and other genetic diseases can still cause death before adulthood. Identify the significance of the symptoms of Huntington disease not usually appearing until later in a person's life.
8. Explain why your reproductive organs are usually shielded with lead sheets during an X-ray.

Use the following information to answer question 9.

Sickle cell anemia is an autosomal recessive genetic disease. The impaired gene causes red blood cells to be produced that are shrunken sickle shapes rather than the normal round disc shapes. These deformed red blood cells can block narrow blood vessels. People with two recessive sickle cell alleles become very ill and often die while they are very young. Most people who suffer from sickle cell anemia or carry the recessive gene tend to be from areas affected by malaria, which is a deadly disease carried by mosquitoes. The malaria parasite reproduces inside a person's red blood cells. People who are carriers of one of the mutated sickle cell anemia alleles actually have an advantage over non-carriers—they are resistant to malaria. This accounts for the fact that this allele is more common in people from areas affected by malaria.

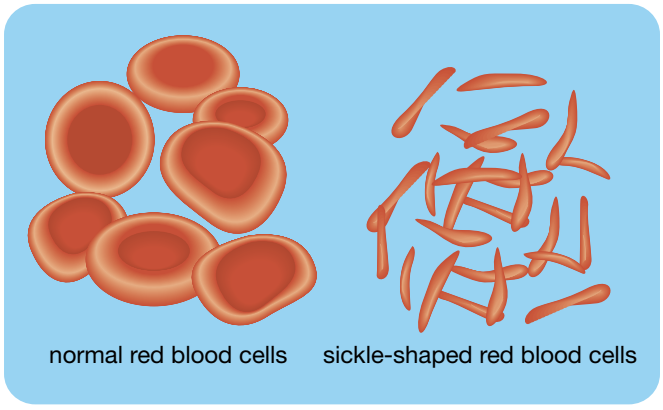


Figure A2.19: The distribution of malaria prior to mosquito control programs is highlighted on this map by green shading.

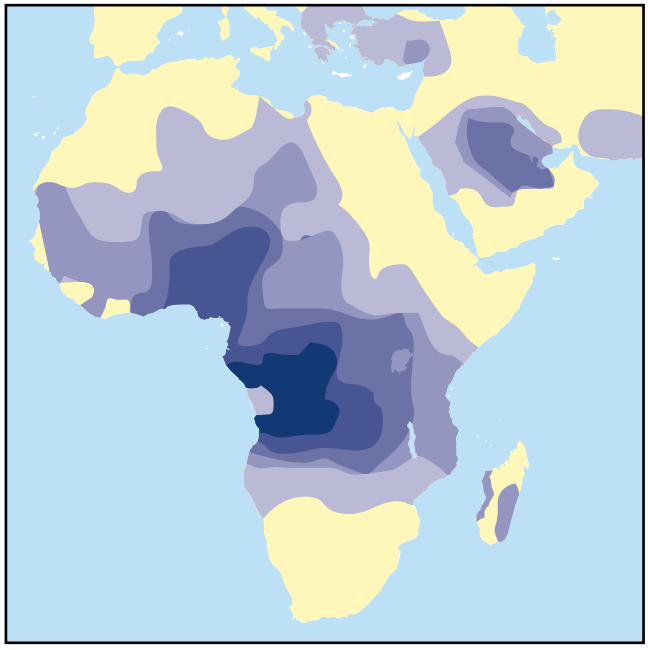


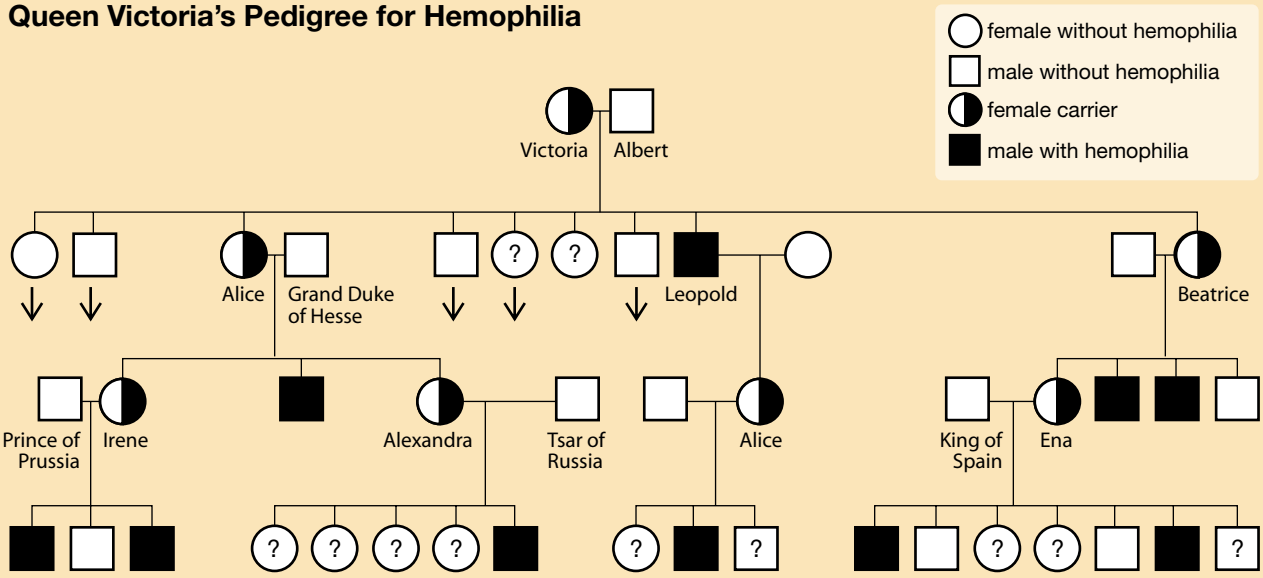
Figure A2.20: This map shows the distribution of sickle cell disease. The darker the shade of purple, the greater the percentage of people who have the disease.

9. **a.** Use a Punnett square to show the probable results of the cross between two people who are heterozygous for the sickle cell trait ($Ss \times Ss$) and are malaria resistant.
   b. List the possible genotypes of the children from this couple.
   c. State the probability of a child of this couple being resistant to malaria.
   d. State the probability of a child of this couple developing sickle cell anemia.
   e. Explain why the sickle cell anemia allele is more common in areas infected with malaria.

Use the following information to answer question 10.

Queen Victoria of England was a carrier of the sex-linked genetic disease called hemophilia. Victoria had many children, but only one of them developed hemophilia. Several of her children married into other European royal families and passed on Victoria's hemophilia allele. The most famous case of hemophilia was Victoria's great-grandson Alexei, the heir to the Russian throne. The controversial historical figure Rasputin gained influence with the Russian royal family by claiming to be able to heal Alexis' hemophilia.

Queen Victoria's Pedigree for Hemophilia



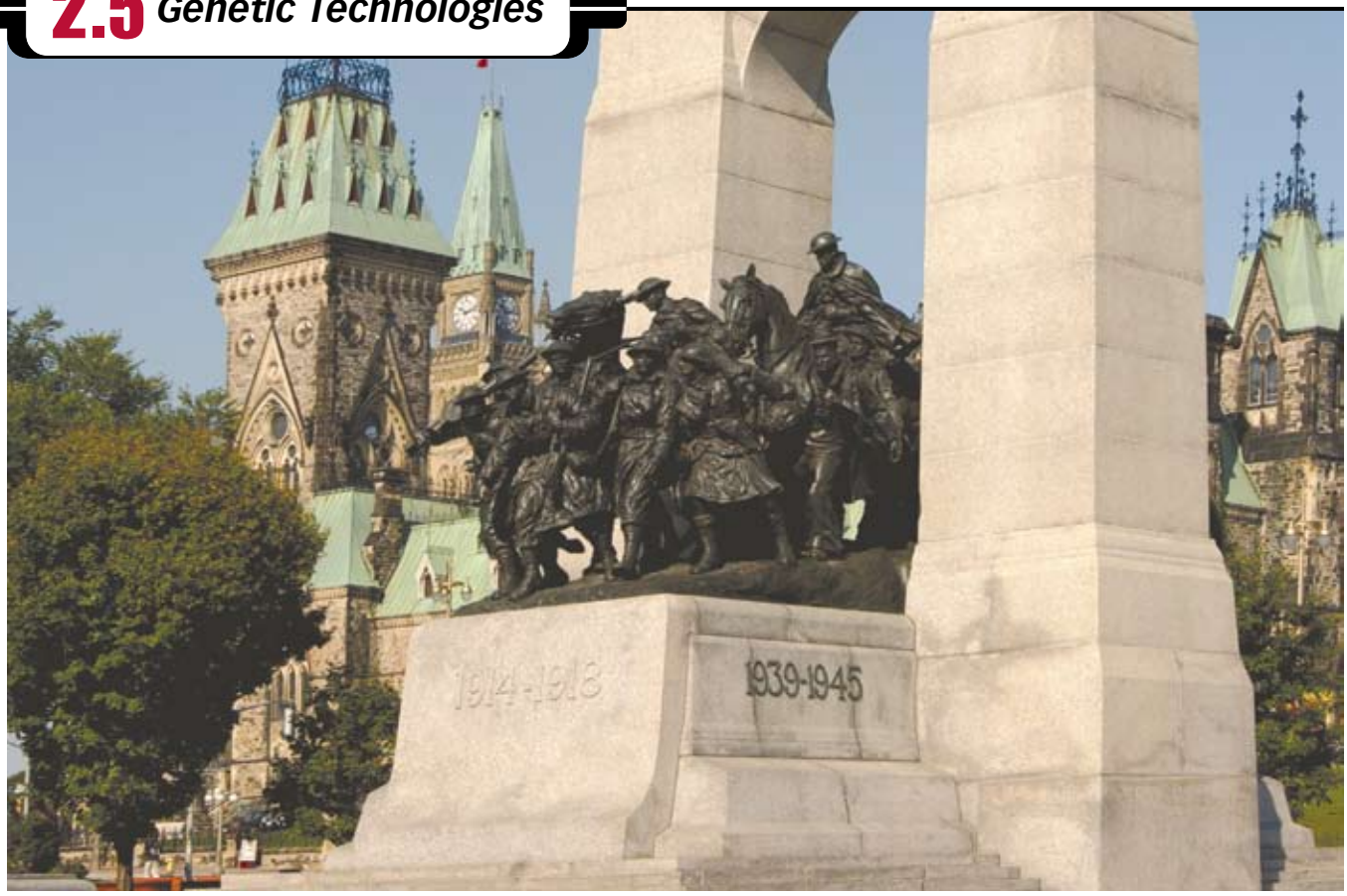
10. Study the pedigree of Queen Victoria's descendants. Then answer questions 10.a. to 10.c.

a. The royal family's ancestry is well documented. There is no history of hemophilia in any of Queen Victoria's ancestors. Explain how you think the hemophilia defect appeared in her genes.

b. Explain why the individuals with hemophilia in this pedigree are all males.

c. Why are question marks written on some of Queen Victoria's female descendants?

2.5 Genetic Technologies



In 2000, the remains of an unidentified Canadian soldier who died during the First World War (1914–1918) were moved from their burial place near Vimy Ridge in France to a special tomb in front of Ottawa's National War Museum. This symbolic "Tomb of The Unknown Soldier" was created to honour the thousands of Canadians who have died in battle and, in particular, those who died without being identified or found.

Methods used by militaries to identify their dead have changed with advances in technology. You may be familiar with the term *dog tag*, which refers to an identification number engraved on a small metal plate that soldiers wear around their necks. Metal dog tags were first used in World War I because, unlike human bodies, they could withstand the force from some bomb blasts and gunfire. Identification tags can be collected after a battle and used to trace which soldiers were killed in an attack. However, as you can imagine, there are many variables in using identification tags as a way to identify troops—tags may fall off, become buried in debris, and be taken by other people at a battle site.

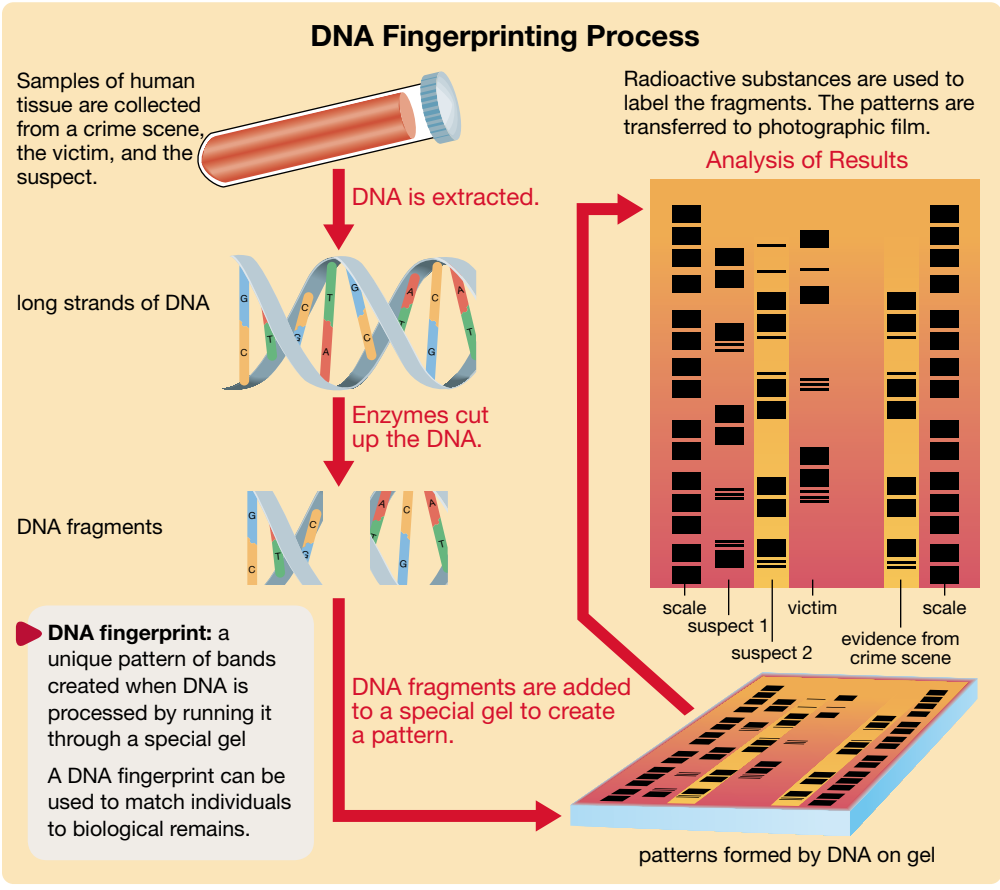
Until recently, an external examination of the body was the most efficient identification method available. The Canadian military has started using DNA obtained by taking a blood or saliva sample from new recruits as a way to store soldiers' biological information for use in identification. Every cell of a person's body contains DNA. When the DNA is extracted and processed it forms a pattern unique to each individual, so examining a person's DNA can be an accurate way of identification. Using libraries of DNA patterns can be a reliable way of keeping track of military personnel, making the possibility of another "Unknown Soldier" a thing of the past.

Advances in the understanding of genetics and the use of genetic technologies have created a scientific revolution. Genetic technologies are being used to identify, treat, and prevent hereditary diseases; to develop new medicines; to solve crimes; to identify individuals such as unknown soldiers; and even to re-design organisms. Although the science of molecular genetics is just over 50 years old, these uses have already made significant impacts on society. Some people worry about the safety and ethics of using these technologies. In this lesson you will learn about and research some genetic technologies. From several perspectives you will be asked to look at issues created with the use of genetic technology. You will also explore the potential ethical implications of using these technologies by performing a risk-benefit analysis of a genetic technology and making a decision about the extent of its use.

DNA Fingerprinting

Analyzing DNA has become a useful and popular tool in forensic investigations. Using DNA testing to solve a crime or to prove a person's innocence has been the subject of many books, popular television shows, and movies. This technology works because of the uniqueness of each person's DNA. In order to identify a person, strands of DNA are isolated from that person's cells and cut into smaller fragments. Then these DNA fragments are separated as they move through a special gel placed within an electric field. As DNA fragments are pulled through the gel by the charged ends of the field, a pattern of bands form. The DNA pattern that appears on the gel for a tested individual is as unique as the swirling patterns of an individual's fingerprints. That's why this process has been called DNA profiling or **DNA fingerprinting**. Identical twins or triplets have the same DNA fingerprints. A DNA fingerprint can be analyzed and compared to DNA collected on a battlefield or at a crime scene, matching evidence to soldiers' remains or to suspects. Similar processes are also used to determine whether people are genetic relatives or to carry out wildlife forensics.

The uniqueness of a DNA fingerprint and the information gained from a person's DNA can be stored in a computerized database. In most countries, only criminals have their DNA profiles stored in computerized DNA databanks. Some people would like to see everyone's DNA stored in a DNA bank. A more complete databank of DNA could be very useful, since a computer can quickly search the stored information. As a result, police would have a better chance of matching evidence from a crime scene that contains DNA—cells, tissue, hair, and saliva—to individuals in the database.



Practice

45. Health benefits provided by employers and health-insurance companies help pay an employee's wages when the employee becomes ill. Explain how a genetic test could be used against a prospective employee or someone applying for health insurance.

46. Genetic information—including the identification of genes that make it more likely to develop certain diseases—can be revealed about an unborn baby by performing an amniocentesis. Describe one risk and one benefit related to the use of genetic test results from an unborn baby.

47. DNA for testing can be collected from a small amount of blood, hair, saliva, and other body fluids. Do you think authorities have the right to collect samples and perform DNA profiling on an individual without this person's permission? Do you think that an individual has the right to refuse to provide a DNA sample for authorities? Once a DNA profile or a genetic screen has been performed, is it possible to keep the results private? Explain your answers.

Transgenics

The mice in Figure A2.21 are glowing because some of the genes they possess have been altered to produce a unique protein with the ability to glow when exposed to ultraviolet light. Jellyfish produce a protein that enables them to glow in certain light, and scientists have isolated this jellyfish gene and then used a modified virus to insert it into the DNA of a mouse embryo. When the mouse embryo develops, each cell has the instructions to make the luminescent jellyfish protein to create a mouse with the ability to glow.

Figure A2.21

The process of intentionally altering the genetic traits of an organism is called genetic modification. As you learned earlier, genetic modification can be done through traditional selective breeding within a species by cross-breeding between closely related species. However, it is much faster to transfer the isolated genes from one species into another species in a process called **transgenics**. People often use the terms **genetic engineering** and transgenics interchangeably, but genetic engineering is a more general term, which includes technology that is hundreds of years old. The result of transgenics is called a **genetically modified organism** or **GMO**.

- **transgenics:** a type of genetic modification in which the gene or genes from one species are transferred and spliced into the DNA of another species
- **genetic engineering:** the modification of genetic material through the actions of people, including selective breeding and modern techniques outside the normal reproductive process of organisms
- **genetically modified organism (GMO):** an organism whose genetic material has been deliberately altered through transgenics

Imagine having the ability to combine the traits from one organism with the traits of another organism. This technology is used to create new foods, medicines, or materials with the potential to increase crop yields, improve health, cure diseases, and produce new products. Many modifications to organisms are being made by genetic engineering, including crop plants containing genes from other organisms that naturally produce their own pesticides. Another modification is bacteria containing a human gene capable of producing insulin required by diabetics.

Scientists are also using this technology to develop pigs with human genes that produce the necessary antigens to make pig organs more compatible with humans and, therefore, more useful for organ transplants. Some researchers are developing goats with genes associated with the silk a spider produces so that goat milk has strands of very strong spider silk for making rope.

The ability to alter organisms with transgenics has an almost limitless number of possible applications. Many industries and companies are interested in this technology, and there is a huge potential to produce both useful and novel inventions that can be sold to make a profit. For example, if the glowing jellyfish gene is combined with an evergreen tree, the inventors could sell glowing Christmas trees that don't need strings of lights.

Opinions on genetic technologies come from many perspectives. From an ethical perspective, some people may question creating genetically modified organisms because they have concerns about harming living organisms. Other people are in favour of transgenics from an economic perspective because the products of this technology have proven to be valuable.

Opposition to the use of this technology has also come from a scientific perspective because some people fear that genetically modified organisms can produce unexpected effects on ecosystems if they are released accidentally. For example, there is a concern that herbicide-tolerant canola may cross-pollinate with related weeds to produce weeds that are herbicide-tolerant. One type of corn has been genetically modified to produce a pesticide. A scientific study has indicated that this corn unfortunately caused the death of monarch butterfly caterpillars.

Many people are opposed to transgenics because they are afraid that some scientists may use genetic engineering to create monstrous creatures like the chimera. The chimera is a mythical beast made from the parts of several different animals. These genetically modified organisms could endure lives of suffering because of genetic experiments. Even more fearful is the possibility that these transgenic organisms could become dangerous either by accident or on purpose. For example, disease-causing organisms could be engineered to become more infectious and deadly by combining the traits of two or more pathogens and then used as a weapon. Genetically modified disease-causing organisms designed to infect people and make them sick or kill them are called **bioweapons**.

People who have concerns or fears about transgenics often refer to works of science fiction where the use of technology goes horribly wrong. Examples include *Frankenstein* and *Jurassic Park*. Foods that have been genetically modified are sometimes even called "Frankenfoods" by people fearful of this technology.

bioweapons: genetically modified disease-causing organisms designed to infect people and either make them sick or kill them

Practice

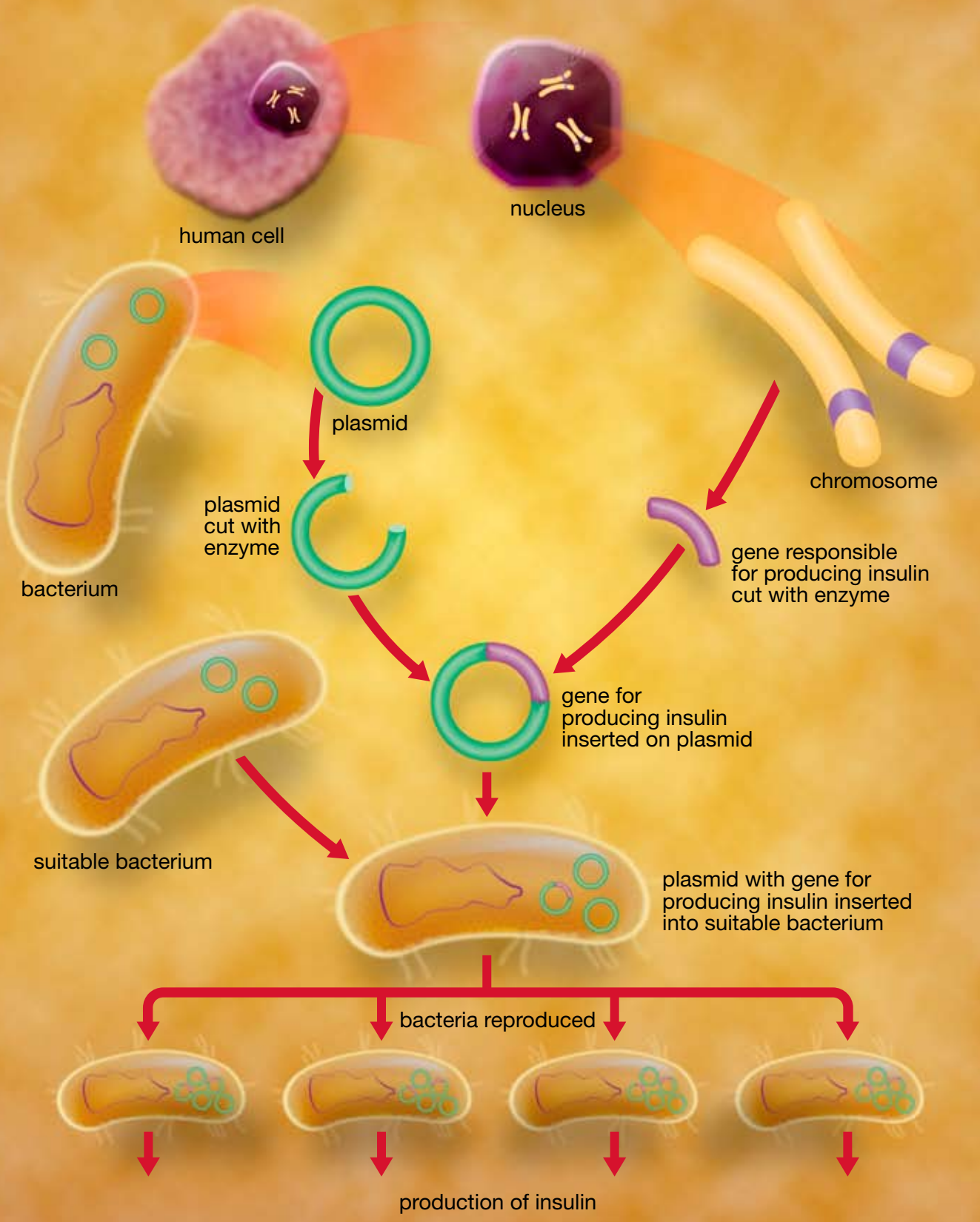
48. At the time this textbook was written, Canada did not require manufacturers to indicate ingredients on their food labels that originate from genetically modified organisms. European Union countries do require such labels. Genetically modified (GM) foods are almost impossible to distinguish from non-genetically modified foods because they usually look the same. Herbicide-resistant GM versions of corn, canola, flax, soybeans, sugarbeets, and wheat are grown in Canada. You have most likely eaten some of these genetically modified foods. Do you think food manufacturers in Canada should be required to indicate genetically modified foods in their products? Explain your answer.

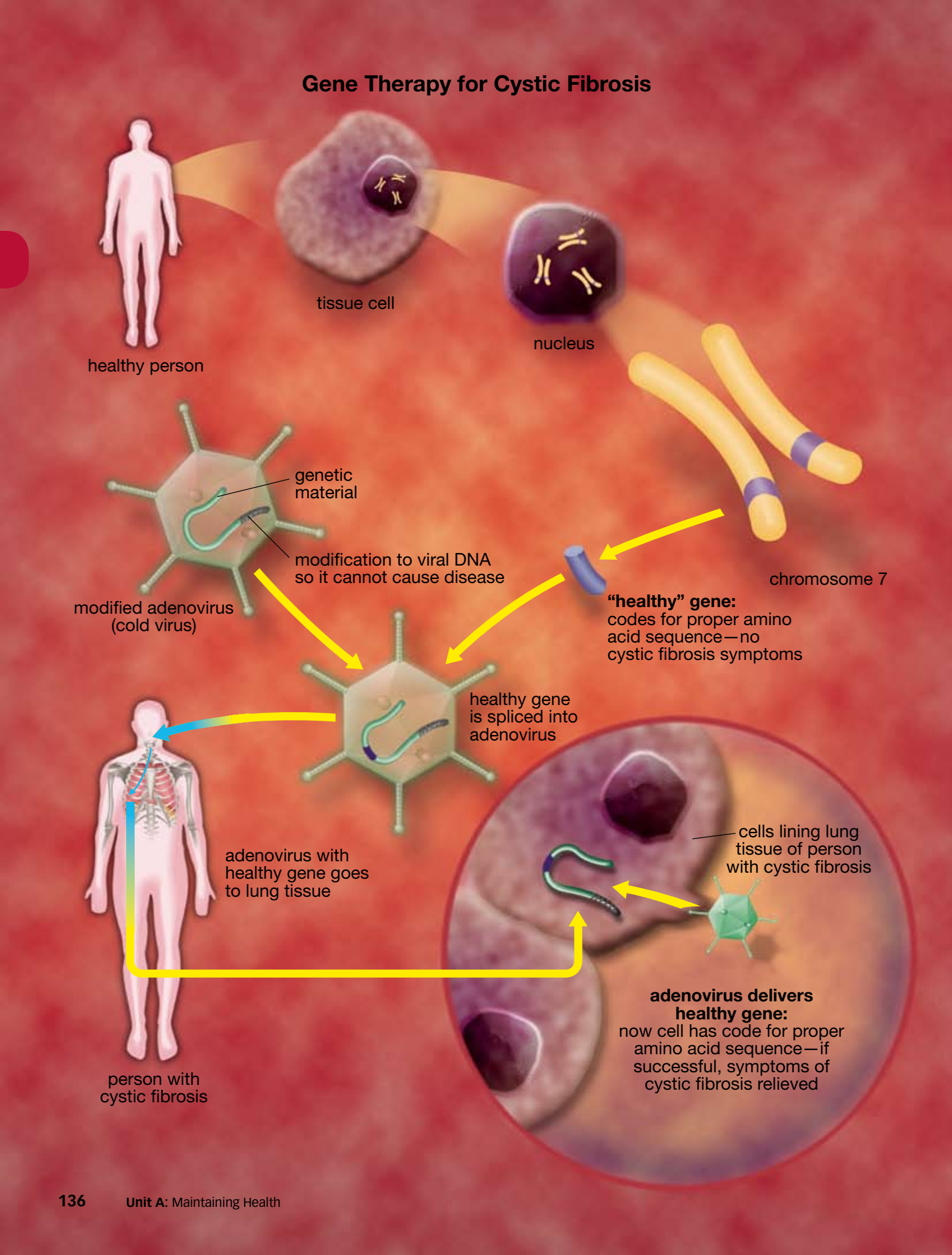
49. A research facility has produced a mouse that does not produce hair and does not initiate an immune response that would reject the ear grafted on its back. The ear grown on a mouse could be used to replace an ear that a person has lost due to an accident. Transgenics could be used to insert human genes into mice, pigs, or other animals to make them even more suited to growing organs that would not be rejected when transplanted into humans. From an ethical perspective, do you think transgenics should be used to create animals with organs compatible for transplanting into humans? Explain your answer.

Applying Transgenics—Medicines and Gene Therapy

Scientists have begun using transgenics to produce medicines instead of having to chemically manufacture medicines or have them collected and extracted from plant or animal parts. Extraction from animal parts is a lengthy process that requires a large input of animal tissue to yield relatively little product. The extraction process requires the use of many noxious chemicals, and the insulin extracted from animals can trigger allergic reactions in some patients.

Producing Insulin Through Transgenics




Gene Therapy for Cystic Fibrosis
healthy person
tissue cell
nucleus
chromosome 7
genetic material
modification to viral DNA so it cannot cause disease
modified adenovirus (cold virus)
"healthy" gene: codes for proper amino acid sequence—no cystic fibrosis symptoms
healthy gene is spliced into adenovirus
adenovirus with healthy gene goes to lung tissue
person with cystic fibrosis
cells lining lung tissue of person with cystic fibrosis
adenovirus delivers healthy gene: now cell has code for proper amino acid sequence—if successful, symptoms of cystic fibrosis relieved

Scientists can now isolate the gene in a healthy human that produces a substance lacking in other people, such as the gene that produces insulin. Enzymes are used to cut a sample of the healthy individual's DNA into pieces. The segment of DNA with the needed gene is isolated, and the gene is inserted or spliced into a plasmid removed from bacteria. DNA containing the genes from two or more organisms—such as combined plasmid with inserted human DNA—is called **recombinant DNA**. The recombinant plasmid is put into a bacterium and huge amounts of this new bacterium are grown. This creates a strain of bacteria that produce insulin. The insulin can be collected and given to people who have diabetes.

- **recombinant DNA:** DNA containing the genes spliced from two or more organisms
- **gene therapy:** the technique of using a vector, such as a virus, to repair or replace defective genes in the treatment and possible cure of genetic diseases

Genetic diseases are difficult to cure because they are caused by a defective copy or copies of an allele present in every cell of a person's body. Some people with genetic diseases can be treated by being given a product that their bodies cannot produce on its own. For example, people with hemophilia cannot produce a type of blood-clotting protein, so they have to receive several injections of clotting proteins per week. Scientists are working on ways to repair or replace non-functioning genes so that genetic diseases can be treated more effectively or even cured. Using genes instead of drugs to treat or cure a disease is called **gene therapy**.

Gene therapy works by identifying and isolating a desired gene from one individual and using it to replace a non-functional gene in another individual. In order for the isolated therapeutic gene to be effective, it must get spliced into the DNA of the person with the non-functional gene. A gene cannot be directly inserted into a person's cells, so one way to insert the therapeutic gene into cells that need to produce the missing protein is to use a vector, such as a virus. Scientists have taken advantage of the way that viruses deliver their genes when they infect cells. The disease-causing genes of the virus are removed and the therapeutic gene is spliced into viral DNA. Patients are infected with many of the altered viruses. Each virus injects the recombinant DNA—containing the therapeutic gene—into a patient's cell to allow the cell to produce the missing or defective protein.

"Bubble Baby" Cured Using Gene Therapy

Figure A2.22 shows a small girl named Salsabil, who had a mutation to the one gene that is responsible for producing an essential enzyme called adenosine deaminase, or ADA. Salsabil did not have a trace of the ADA enzyme in her body because the mutated gene was defective. Since ADA is responsible for producing T-cells and B-cells, Salsabil had virtually no immune system and had to live the first seven months of her life in a plastic bubble to protect her from pathogens. This is why this illness—severe combined immunodeficiency or SCID—is often called "bubble baby syndrome" in the media.

Figure A2.22: Dr. Shimon Slavin holds a healthy Salsabil prior to her second birthday.

Children with SCID used to be treated with injections of the ADA enzyme every two days or they received a transplant of healthy bone marrow from a compatible donor. If neither treatment was possible, the only alternative was for the children to live their lives in artificial, germ-free environments.

In Salsabil's case a team of doctors and medical researchers, led by Dr. Shimon Slavin, were able to use gene therapy. A sample of Salsabil's bone marrow cells were extracted and mixed with a genetically engineered virus containing a healthy copy of the defective gene. The virus injected the human gene directly into the nucleus of the bone marrow cells. Before the healthy bone marrow cells were transfused back into Salsabil, the medical team subjected her to a mild form of chemotherapy to suppress her defective bone marrow cells. When the healthy cells were introduced to Salsabil's body, they had not been subjected to the chemotherapy and had an advantage—they began to take over and grow rapidly. Within months, Salsabil had T-cells and B-cells working together to produce antibodies. A year later, Salsabil returned to her family. She was effectively cured of SCID.

Practice

50. Describe some concerns that arise from using viruses to carry and insert therapeutic genes into patients.

51. The use of gene therapy is currently focused on treating and curing genetic diseases. As more genes become identified and studied and this technology becomes more advanced and accessible, some people who can afford this technology might seek to use it to alter genes that control traits—such as height, intelligence, or athletic ability—other than those causing disease. People might use this genetic technique to insert desirable genes either into themselves or into embryos before they begin to develop. Describe some risks associated with using gene therapy techniques for goals other than treating and curing diseases.

52. Insulin can now be produced by genetically engineered bacteria. Before these bacteria were approved, insulin could only be obtained through extractions from the pancreases of pigs or other livestock. List some benefits of using genetically engineered bacteria instead of animal glands.

53. Choose one of the following problems to design an experimental procedure to investigate a characteristic of a genetically modified organism. In your experimental design, list the steps you would take to carry out your experiment. State the manipulated variable, responding variable, and at least three controlled variables.

a. A genetic engineering company has produced a genetically modified variety of onions. The company isolated the protein that makes our eyes water when onions are cut, and people who work for the company believe that they can make the gene that produces the protein non-functional. This genetically modified (GM) onion will not make people's eyes tear when they cut the onion. Some researchers are worried that removing this gene will affect the taste of the new GM variety. Design an experiment to determine whether the onion's flavour has been affected by removing the eye-watering gene.

b. Genetic engineers have isolated a gene from a cold-water fish called a flounder. This gene produces a protein that acts like antifreeze—it prevents the fish from freezing in the icy waters where it lives. Researchers have inserted the gene responsible for producing the antifreeze protein into a tomato plant's genetic instructions to make a new variety of tomatoes more resistant to frost.

If the new variety of tomatoes proves to be effective and popular, then fewer tomatoes will spoil during shipping. Design an experiment to determine whether the new tomato variety is more resistant to frost than non-modified varieties.

c. Agricultural scientists have used the genetic modification technology of gene splicing to insert a gene into a canola plant that makes the canola more resistant to strong chemical herbicides. A crop of GM canola can then be sprayed with a strong chemical herbicide to kill competing weeds and leave the canola crop unaffected. With no competition from weeds, the GM canola should grow more easily and produce a greater yield of canola oil than a non-engineered crop. Design an experiment to determine whether the herbicide-resistant canola crop produces a greater yield than a non-modified crop.

Genetic Techniques Used in Molecular Biology

The transfer of DNA fragments between bacteria species is at the heart of much current work in molecular biology. As shown in Figure A2.23, the data collection for this research involves specialized equipment and carefully developed techniques. In the next activity you can learn more about how this research is conducted.

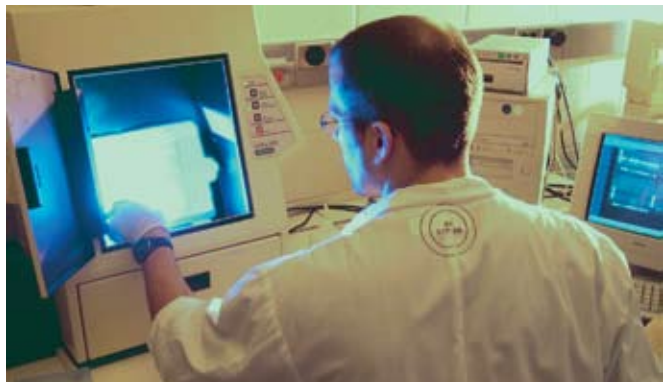


Figure A2.23: A researcher collects data using a fluorescent dye to determine the quantity of specific proteins from a DNA sample.

Investigation

Risk-Benefit Analysis—Genetically Modified Foods

Purpose

You will use information presented in this chapter and from the Internet to gather data regarding the risks and benefits of genetically modified foods. You will develop a position on this issue and then defend this position in light of information presented by other students.

Science Skills

- ✓ Initiating and Planning
- ✓ Performing and Recording
- ✓ Analyzing and Interpreting
- ✓ Communication and Teamwork

Identify Alternatives/Perspectives

1. To what extent should genetically modified (GM) foods be developed and used? Begin to brainstorm alternative solutions to this question. One approach is to consider the question from as many points of view as possible. Think of several different groups or individuals who have a particular view or interest on the use of genetically modified foods. Create a table with "Stakeholder" at the top of one column and "Point of View/Perspective" at the top of the other column. Complete the table by listing at least three stakeholders and their viewpoints.

Research the Issue

2. Conduct research to collect and assess information for all the perspectives of stakeholders you have identified. Assemble the relevant information as points on a page. People use a variety of terms when referring to genetically modified foods. For a more effective Internet search, you should perform several searches using the many variations on terms in your search engine: *genetically modified foods*; *GM foods*; *genetically engineered foods*; *GE foods*; *biotech foods*; *biotechnology*; *genetically modified organisms*; *GMO*; and other examples.

Analyze the Issue

3. **a.** Analyze the results of your research by concisely organizing your findings in a second table, with "Risks" at the top of one column and "Benefits" at the top of the other.
   b. Review the risks and benefits. How would each stakeholder react to the entries? Record the reactions of three of your stakeholders to the data on your Risk/Benefit table.

Take a Stand and Defend Your Position

4. To what extent should genetically modified (GM) foods be used? What is your position? Take a clear position on this issue by writing a few concise paragraphs. Your position should be supported by the body of research and should indicate that you have considered the question from more than one viewpoint.

Evaluation

5. It is very helpful at this stage to share your findings with other students. How do their points of view differ from yours? Are the arguments made to support these views consistent with the information that you researched? Did other students find additional information that was unknown to you? How has your position changed since you started? If you had to make this decision again, what would you have done differently?

   Write a few concise paragraphs to evaluate your position and the process you used to develop this point of view. Your response should indicate that you have considered the positions of other students and that these alternative viewpoints have been addressed.

2.5 Summary

Each person's DNA is unique, and the technology of DNA fingerprinting can be used for identification purposes.

People can genetically alter the genetic traits of an organism. This genetic modification can be done using selective breeding within a species or a closely related species. When genetic modification is done by inserting a gene or genes from one species into another species, it is called transgenics. Transgenics can be used to produce genetically modified organisms, or GMOs, used for new kinds of foods, medicines, or materials.

Some people oppose the production of genetically modified organisms. Concern about the development of genetically engineered organisms tends to focus on issues regarding possible dangerous or as yet unknown effects on people or the ecology. Some people feel that it is cruel to make experimental organisms, or they disagree with changing organisms at all. Others fear that genetic engineering technology will be used to make bioweapons. Opposition can also be based on moral or religious reasons.

Transgenics is accomplished by making recombinant DNA, which is a combination of genes from two or more species spliced together.

Recombinant plasmids are used in bacteria to produce large amounts of needed medical enzymes such as insulin.

Repairing defective genes by inserting a non-defective copy into a person's DNA is called gene therapy. Therapeutic genes are inserted with the help of virus vectors that have had their disease-causing genes removed. The viruses deliver the therapeutic gene to cells in patients.

2.5 Questions

Knowledge

1. Match the following terms with the example that best describes each term.

- transgenics
- genetic modification
- DNA fingerprinting
- recombinant DNA
- gene therapy

a. A farmer uses a plastic bag to collect pollen from his fastest-growing corn plants and then sprinkles some pollen on the corn silk of his most disease-resistant corn plants. He collects the seeds produced from this cross-pollination and grows his next crop from these seeds.

b. Enzymes are used to cut up DNA left at a crime scene, and then the DNA is run through a gel. The distinctive pattern of bands produced is used to compare with the patterns of suspects in the crime.

c. A modified virus is used to deliver a non-defective version of the gene that causes cystic fibrosis in body cells.

d. A researcher uses enzymes to cut some human DNA into smaller pieces and then uses different enzymes to splice the DNA into a bacterial plasmid. The new DNA is a combination of bacterial DNA and human DNA.

e. A gene from the bacterium *Bacillus thuringiensis* (Bt) produces a protein with insecticidal properties. The bacterial gene is isolated and spliced into the DNA of a cotton plant. When the cotton plant is grown, it produces the bacterial insecticide.

2. List some potential advantages of genetically modified plant crops.

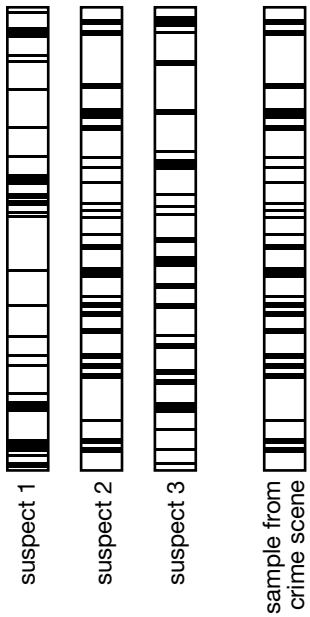
3. Describe one possible risk of applying transgenics to produce a new type of organism.

4. Explain why virus vectors need to be used in gene therapy.

Applying Concepts

5. A sample of biological material was left at a crime scene. The DNA from this sample was isolated and a DNA fingerprint was created. The three crime suspects all volunteered to let their DNA be taken, and a DNA fingerprint was created for each person. Compare the unique banding pattern produced for each suspect to identify the suspect who was at the crime scene. This information can be seen in "DNA Fingerprint Patterns."
6. Explain why a person who receives gene therapy will not pass on to her offspring the repaired gene that has been inserted into her cells by a virus.
7. The gene that produces a blood-clotting factor that some people who have hemophilia lack has been isolated. Explain the steps used to develop a strain of genetically engineered bacteria that produce large amounts of this factor to treat people with hemophilia.
8. Most of the citizens of Iceland have volunteered to have their genetic information collected and compiled in an electronic database. The genetic make-up of Iceland's population has changed little since the Vikings colonized the island in the ninth century. This history makes it easier for researchers to identify gene mutations that may be associated with diseases. Describe one risk and one benefit of a nation possessing a gene bank for its citizens.

DNA Fingerprint Patterns



Chapter 2 Summary

In this chapter you have examined the structure and function of DNA and have applied your understanding of the mechanisms of DNA inheritance to predict the probability of offspring inheriting traits caused by a single gene. You have also seen how mutations can affect the functioning of DNA and how certain diseases can arise from inherited mutations rather than from environmental factors. You used tools, such as a Punnett square and a pedigree chart, to predict and trace the inheritance of traits in individuals within a family. In Lesson 2.5 you examined the use of genetic technologies and looked at the ethical implications related to their use. Throughout the chapter you learned about the contribution of Mendel and other scientists to the field of genetics.

In future units of Science 30, you will learn more about factors that increase the likelihood of mutation. These factors include chemical substances used in the production of commonly used materials—or substances considered to be pollutants—and some forms of radiation within the electromagnetic spectrum.

Summarize Your Learning

In this chapter you learned a number of new biological terms, processes, and theories. It will be much easier for you to recall and apply the information you have learned if you organize it into patterns.

Since the patterns have to be meaningful to you, there are some options about how you can create this summary. Each of the following options is described in "Summarize Your Learning Activities" in the Reference section. Choose one of these options to create a summary of the key concepts and important terms in Chapter 2.

Option 1: Draw a concept map or a web diagram.	**Option 2:** Create a point-form summary.	**Option 3:** Write a story using key terms and concepts.	**Option 4:** Create a colourful poster.	**Option 5:** Build a model.	**Option 6:** Write a script for a skit (a mock news report).

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Chapter 2 Review Questions

Knowledge

1. Describe the differences among the terms *chromosome*, *DNA*, and *gene*.
2. How many chromosomes are found in a human autosomal cell?
3. Compare the processes of mitosis and meiosis.
4. What are the differences between acquired and inherited characteristics?
5. A pea plant with green pea pods is crossed with a pea plant with yellow pea pods. Both plants come from lines of pea plants that have only produced one colour of pod. All offspring from this cross develop green pea pods.
   a. Determine whether the green colour for pea pods is dominant or recessive.
   b. Using letters to represent alleles, write the most likely genotypes for the two parent plants and the genotype of the offspring.
6. Distinguish between the terms *genotype* and *phenotype*.
7. For each of the following DNA nucleotide sequences, write the sequence for the complementary strand and for the chain of amino acids that code for this strand.
   a. ATATACCAGCCGATA
   b. GCATGGTTCATAAGG
   c. CGTATGCCAGTTTAT
   d. GGTTTATGCATTTCT
8. For each of the following amino acid chains, write all the corresponding DNA sequences that could code for that chain.
   a. Methionine-Threonine-Glutamine
   b. Arginine-Lysine-Tryptophan
   c. Serine-Proline-Aspartate
   d. Leucine-Cysteine-Valine
9. Describe the importance of proteins.
10. Compare and contrast a point mutation and a frameshift mutation.
11. List the steps used to create recombinant DNA.

Applying Concepts

12. Draw a series of diagrams to compare and contrast mitosis and meiosis. For each step, use one pair of chromosomes and include labels to describe the process.
13. A rare recessive allele causes a lack of fur pigment in tigers to produce the distinctive "white tiger" phenotype. These animals have sometimes been incorporated into the extravagant stage shows of Las Vegas magicians.
    a. Explain why entertainers who use the white tigers in their shows only want their white tigers to breed with other white tigers or the offspring of white tigers.
    b. State possible problems with the selective inbreeding of closely related white tigers.

14. A dog breeder owns a dog that has just given birth to a litter of puppies. Both the father and the mother were selected from long lines of well-known and recorded pedigrees. One of the puppies has a red fur colouring that the breeder has never seen in any of the puppy's ancestors.
    a. Explain the likely cause of this new trait.
    b. Describe how you can determine if the new colour trait is dominant or recessive. Use Punnett squares to illustrate your answer.
    c. Explain the steps you would take to develop a breed of dogs with this particular trait.
15. The use of antibacterial soaps has become popular. Describe how bacteria can develop a resistance to antibacterial soaps. Include the role of bacterial plasmids in your answer.
16. Draw a series of images that illustrate the process of gene therapy on a patient who has the autosomal recessive disease of cystic fibrosis.

17. A breeder of Labrador retrievers is told that black fur is dominant over yellow fur. The breeder crosses a black Lab with a yellow Lab. When the puppies are born, some are black, some are yellow, and some are chocolate in colour. Evaluate the following statements regarding this Labrador cross. State whether you agree or disagree with each one. For those statements you disagree with, explain why.

a. The gene for coat colour in Labrador retrievers must be controlled by more than one gene or have more than two possible alleles.

b. All the puppies have homozygous genotypes for their coat colour.

c. The female must have bred with both a black Labrador male and a chocolate Labrador male to produce three kinds of coats in her puppies.

d. A pedigree of coat colours is helpful to determine the genotype of the parents and offspring.

18. The allele that produces hairy ears in humans is found on the *Y* chromosome.

a. State which gender is affected by the presence of a gene on the *Y* chromosome.

b. Explain why a Y-linked gene cannot be recessive or dominant.

c. State the probability of a male with hairy ears passing this trait onto his son.

d. State the probability of a male with hairy ears passing this trait onto his daughter.

e. Can a person be a carrier for this trait? Explain your answer.

19. Elliptocytosis is a genetic disorder affecting a protein that influences the cell membrane structure in red blood cells. Red blood cells with the altered protein have an elliptical, or oval, shape when compared to red blood cells containing the unaltered protein.

The pedigree in Figure A2.24 shows how elliptocytosis is inherited within one family.

a. Based on the evidence shown in this pedigree, determine which pattern of inheritance—sex-linked, autosomal recessive, or autosomal dominant—is exhibited by elliptocytosis. Explain how evidence in the pedigree supports your answer.

b. Predict the possible genotypes and phenotypes of offspring in generation V.

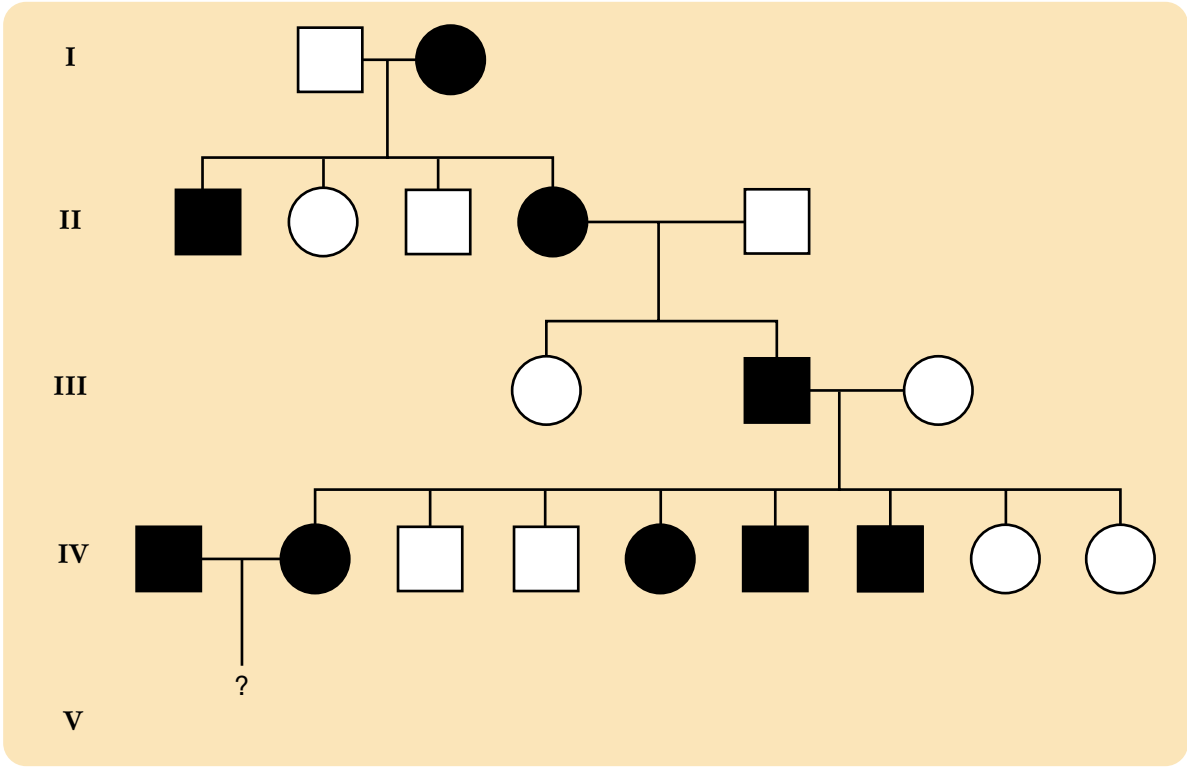


Figure A2.24

Unit A Conclusion

A key theme in Unit A has been the idea of keeping healthy. This unit has examined how your health can be affected by both lifestyle choices and the genes that you inherit. In the first part of the unit you examined how the circulatory and immune systems work together to keep you healthy. You studied the major components of these two systems and considered factors that can affect how they function. A better understanding of these two body systems will help you make healthy choices. In the second chapter of Unit A you looked at the major principles of genetics, and you used these concepts to explain how some traits and diseases can appear more often in some families.

Throughout the unit you investigated and evaluated technologies used to explore, maintain, repair, and assist our bodies. Genetic discoveries and—in particular—the use of genetic technologies can be controversial, and you evaluated their use and made decisions about the extent they should or could be used.

The theme of maintaining health will still be relevant in upcoming units as you study the health effects of radiation and of chemicals released into the environment.

Career Profile

Research Scientist—Pulmonary Medicine

Malcolm King's father was the first person on his reserve to graduate from university. His example made Malcolm seriously consider following in his dad's footsteps to become a high school teacher. Malcolm took that dream down a different path; and today, he teaches pulmonary medicine at the University of Alberta in Edmonton.

Malcolm is not only a professor, but a prominent research scientist. His main area of interest is mucus rheology—the study of the flow of mucus in lungs and other organs. His research focuses on developing treatments for diseases that affect the ability of people to breathe, such as asthma, bronchitis, and cystic fibrosis. Two of the treatments he has developed for chronic lung disease have been patented. Malcolm admits that the patents are two of his greatest scientific accomplishments so far.

Malcolm's passion for science began as a young man. He recalls, "Mr. E.R.S. Hall, my high school chemistry teacher, made chemistry interesting, especially [the] lab experiments." After high school, at age 17, Malcolm moved to Hamilton to attend McMaster University, where he obtained a Bachelor of Science degree in chemistry. He then moved to Montreal, where he obtained a PhD in Polymer Chemistry from McGill University in 1973.

Malcolm knows that school can be tough. For him, the hardest part was "setting priorities for work and study." But he got through it by staying focused on his long-term goals. As someone who knows, he encourages students to "stick with it—there are some really interesting opportunities ahead for Native people." According to Malcolm, "education is an integral part of economic well-being. We need to understand how the economy works if we are to control our destiny."

As the project leader for the University of Alberta's Aboriginal Health Training Initiative, Malcolm aims at increasing the number of Aboriginal students in medicine, dentistry, and related professions. He believes that "you don't have to give up your Native identity when you take up science and engineering. "In fact, it can only help with the well-being of your community."

Unit A Review Questions

1. Match each of the following definitions relating to heart structures to its correct term.

- pulmonary artery
- vena cava
- pulmonary vein
- ventricle
- septum
- atrium
- heart valve
- aorta

a. one of the heart's upper chambers that receives blood returning to the heart

b. one of the heart's lower chambers that pumps blood from the heart

c. a thick wall of muscle that divides the left and right sides of the heart

d. a thin flap of tissue inside the heart that regulates the direction of blood flow within the heart by preventing the backflow of blood

e. the large vein that collects oxygen-poor and carbon dioxide-rich blood from the upper (superior) body and lower (inferior) body

f. the vessel that carries blood away from the heart and toward the lungs

g. the vessel through which oxygen-rich blood flows toward the heart

h. the body's largest artery

2. Record the following table in your notebook. Leave enough room for your responses. Sketch a cross section of the three main types of blood vessels to indicate the relative differences in size and structure between these three types of vessels. Below each sketch, indicate the role of that vessel in the circulatory system.

Artery	Vein	Capillary
Sketch	**Sketch**	**Sketch**
Role	**Role**	**Role**

3. Match each blood component listed on the left with its correct function.

- red blood cell
- white blood cell
- platelet
- sample of plasma

a. a cell that defends the body against disease

b. a yellowish liquid like the broth of a soup made up mostly of water with substances dissolved or floating in it

c. a blood cell that contains hemoglobin and transports oxygen from the lungs to the body's cells

d. a very tiny cell fragment that helps seal skin cuts by initiating clot formation

4. State similarities and differences between the following three disorders of the circulatory system—angina, a heart attack, and a stroke.

5. The different types of white blood cells each have a specific role that can be compared to the role of people and objects associated with a hockey game. Complete the following table by matching each type of white blood cell to a person or object from a hockey game.

a. macrophage
b. memory B- and T-cells
c. B-cells
d. helper T-cells
e. suppressor T-cells
f. antigen

Person or Object and Role	Type of White Blood Cell
Coach—co-ordinates and directs team's moves and analyzes other team's plays	
Offensive players—try to keep opposing team's players and puck on opposition side	
Coach's playbook—keeps records of strategies used to win games	
Referee—calls an end to game	
Jersey—makes a distinction between players	
Defensive players—respond to opposing players who have broken through their defensive line	

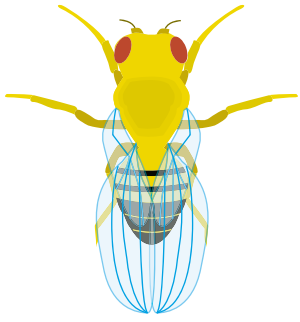
6. Match each term relating to the structure of genetic material with its corresponding definition.

- DNA base triplet
- chromosome
- gene
- karyotype
- amino acid
- protein
- gamete
- DNA

a. all chromosomes from one nucleus matched into their respective pairs and arranged from the largest pair to the smallest pair
b. a sex cell, such as a sperm or an egg, produced during meiosis
c. a molecule that forms a twisted-ladder shape
d. a segment of a DNA molecule that carries information resulting in the production of a specific protein
e. one of 20 possible building blocks used to form proteins as determined by the sequence of bases along a DNA molecule
f. the condensed form of all DNA visible when cells are viewed with a microscope
g. a molecule made from a chain of amino acids that serves a variety of functions in the body including transport, communication, and regulation
h. three adjacent nitrogen bases found along a DNA molecule that code for an amino acid to be produced or begin or end the reading of a gene

7. *Drosophila melanogaster* is a commonly studied species of fruit fly having normal-sized wings. *Drosophila* is dominant over a fruit fly having much smaller wings, called vestigial wings. Use the symbol *W* to represent the allele for normal-sized wings and the symbol *w* to represent the allele for vestigial wings.

a. Write the possible genotypes for a fruit fly with wings of a normal size.
b. Write the genotype for a fruit fly with vestigial wings.
c. Write the possible genotype of the gametes for a fruit fly that is heterozygous for the wing-size trait.
d. State the phenotype of a fruit fly with the *ww* genotype.



8. Describe how the listed components of DNA combine to form a DNA molecule.

deoxyribose sugars
phosphate groups
four nitrogen bases

9. Identify the key characteristics in the following genetic diseases.

a. an autosomal dominant disease

b. an autosomal recessive disease

c. a sex-linked disease

10. List the basic steps involved in producing and using a DNA fingerprint to positively identify biological evidence left at a crime scene.

Use the following information to answer question 11.

A baby will occasionally be born with a hole in the septum between the right and left ventricles. This causes the flow of blood through the heart to be disrupted. Septal defects result in a large amount of blood bypassing the lungs as it leaks through the septum. This impairs the efficient delivery of oxygenated blood to the body. A small hole in a baby's septum usually closes up on its own, but larger holes require surgery to be sealed. Babies born with this defect are often referred to as *blue babies* because they have a bluish tinge to their skin.

11. Explain why the skin of a baby with a septal defect might seem bluer than the usual pinkish-red coloured skin of a newborn baby.

Use the following information to help you answer question 12.

Blood types are determined by the presence of antigens on the surface of a red blood cell. One type of antigen is called the Rhesus factor or Rh factor. The production of the Rh antigen is determined by a single gene with two different alleles. The Rh positive (or Rh^+) allele produces the antigen and the Rh negative (or Rh^-) allele does not produce the antigen. The Rh^+ allele is dominant over the Rh^- allele.

12. a. Draw a Punnett square for a cross between a male with the genotype Rh^+/Rh^- and a female with the genotype Rh^-/Rh^-.

b. State the percentage probability—as a percentage or a ratio—that the offspring will have the *Rh* positive blood type.

c. State the percentage probability that the offspring will have the *Rh* negative blood type.

d. State the percentage probability that the offspring will have the *Rh* positive blood type but carry the *Rh* negative blood type allele.

e. Individuals with the *Rh* negative phenotype produce an immune response to the presence of blood cells possessing the *Rh* antigen. Describe the action of the immune system if someone with the *Rh* negative phenotype is given a transfusion of blood containing cells that are *Rh* positive.

13. A patient has a blood sample taken. Describe the possible symptoms a patient will likely experience given each of the following blood test results.

Blood Test Results	Possible Symptoms
a low red blood cell count	
a high white blood cell count	
a very low T-cell count	
a low platelet count	

14. Scientists are using genetic engineering to develop pigs that incorporate human genes. The pigs can then produce human antigens. The purpose of this technology is to make organs from the genetically engineered pigs usable for transplantation into humans.

a. Explain, in general terms, how the process of genetic engineering can be used to place human genes into pigs.

b. List one risk and one benefit of using genetic engineering to produce these modified pigs.

15. Cyclosporin is a drug that suppresses the immune system. It is given to recipients of organ transplants. Cyclosporin acts to reduce the number of white blood cells—particularly the killer T-cells—thereby reducing the number of white blood cells able to attack the transplanted organ. In many cases, recipients have to take immune-suppressant drugs for the rest of their lives to avoid rejecting the new organ.

a. Identify a negative effect that might result from taking an immune-suppressant drug like cyclosporin.

b. Explain why a person who receives an organ transplant from an identical twin would have much less of a chance of having the organ rejected.

16. A karyotype of an individual is completed by using samples of the individual's blood. State what a karyotype distinguishes, and explain its uses.

17. A gardener crossed a white-flowered tulip with a red-flowered tulip. He then collected the seeds from the offspring of this cross and later planted them. The offspring from this cross all grew red flowers.

a. Identify which of the two colours is dominant.

b. State the genotype for offspring of the cross of two parents.

c. When Carter, a gardener allowed the second-generation tulips to self-fertilize, he found that the seeds produced both red tulip and white tulip plants. When Carter counted and compared the differently coloured plants, he found that there were about three red tulips for every single white tulip. Explain the proportion of each colour observed in the most recent generation of plants.

18. Examine the three different point mutations that have occurred in copies of the same DNA sequence. Identify the mutation with the greatest effect on the amino acid chain produced relative to the original strand.

Original DNA sequence: AGGGCGCCGTTATAT

Mutated DNA sequence # 1: CGGGCGCCGTTATAT

Mutated DNA sequence # 2: AGGGCGCCGTAATAT

Mutated DNA sequence # 3: AGGGCGCCGTTATAC

Use the following information to answer question 19.

Tsarina Alexandra of Russia carried the recessive form of the gene causing hemophilia, which is found on the *X* chromosome. Tsar Nicholas II did not have hemophilia.

19. a. Construct a Punnett square to demonstrate the cross of alleles possessed by the Russian Monarch Tsar Nicholas II and his wife, Tsarina Alexandra.

b. State the probability of a male offspring from this cross developing hemophilia.

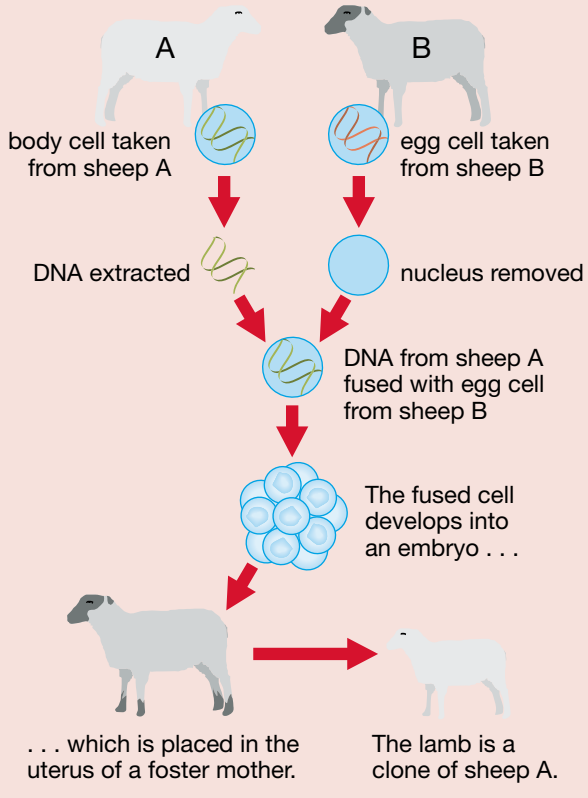
c. Nicholas and Alexandra's only son, Alexei, developed hemophilia, but his four older sisters did not have hemophilia. Explain how it is possible that none of his sisters developed hemophilia.

d. The famous historical figure named Rasputin gained influence with the Russian royal family because he claimed that he could heal Alexei's hemophilia. Use your knowledge of the circulatory system and genetics to explain why it could not have been possible for Rasputin to cure Alexei of hemophilia.

e. The Tsar and his family were assassinated as part of the Russian Revolution of 1917. Years later, a woman appeared and claimed to be Anastasia, one of the daughters of Nicholas and Alexandra. Throughout her life, this woman maintained that she was Anastasia. A DNA sample from a piece of her intestine was removed during an operation and preserved. This sample was compared to DNA from the royal family members, whose bodies were uncovered in 1998. This DNA comparison was used to prove that she was not Anastasia. How can DNA be used to identify members of the same family?

20. Use the following information to answer questions 20.a. to 20.f.

During the process used to create the clone of a sheep, scientists took an egg cell from one sheep and removed its chromosomes. An autosomal cell from a second adult sheep was isolated and placed next to the egg cell. Electricity was used to fuse the two cells together, and they began to divide into a zygote, which was genetically identical to the adult sheep's autosomal cell. The clone zygote was implanted into a third sheep's uterus, where it developed into a baby sheep.



a. Would the egg cell be described as haploid (1n) or diploid (2n) before its chromosomes were removed?

b. Would the autosomal cell from the adult sheep be described as haploid (1n) or diploid (2n)?

c. Would the clone zygote produced from the two fused cells be described as haploid (1n) or diploid (2n)?

d. Explain why the chromosomes needed to be removed from the egg cell to create the clone zygote.

e. Compare and contrast the process of cloning with the process of fertilization.

f. Even though the cloned sheep and the sheep from which the autosomal cell was taken share all the same genes, they may differ slightly. Explain how slight differences such as adult size and behaviour are possible.