

## 2

# Reproduction

When wild salmon hatch from their eggs they grow to market size in 20 to 30 months. AquAdvantage™ salmon, produced at a fish farm in Newfoundland and Labrador, are genetically engineered for more rapid growth. These fish reach market size twice as fast as conventional salmon. Scientists need detailed knowledge of heredity and reproduction to carry out genetic engineering. They also continue to research how human-controlled reproduction can affect the survival of wild salmon populations.

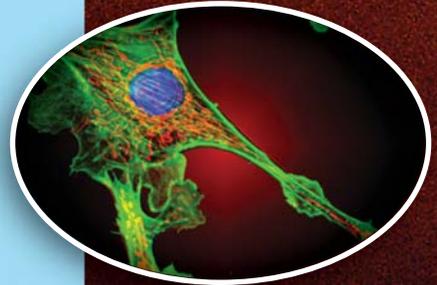


## Key Ideas

4

### The nucleus controls the functions of life.

- 4.1 The Function of the Nucleus within the Cell
- 4.2 Mutation



5

### Mitosis is the basis of asexual reproduction.

- 5.1 The Cell Cycle and Mitosis
- 5.2 Asexual Reproduction



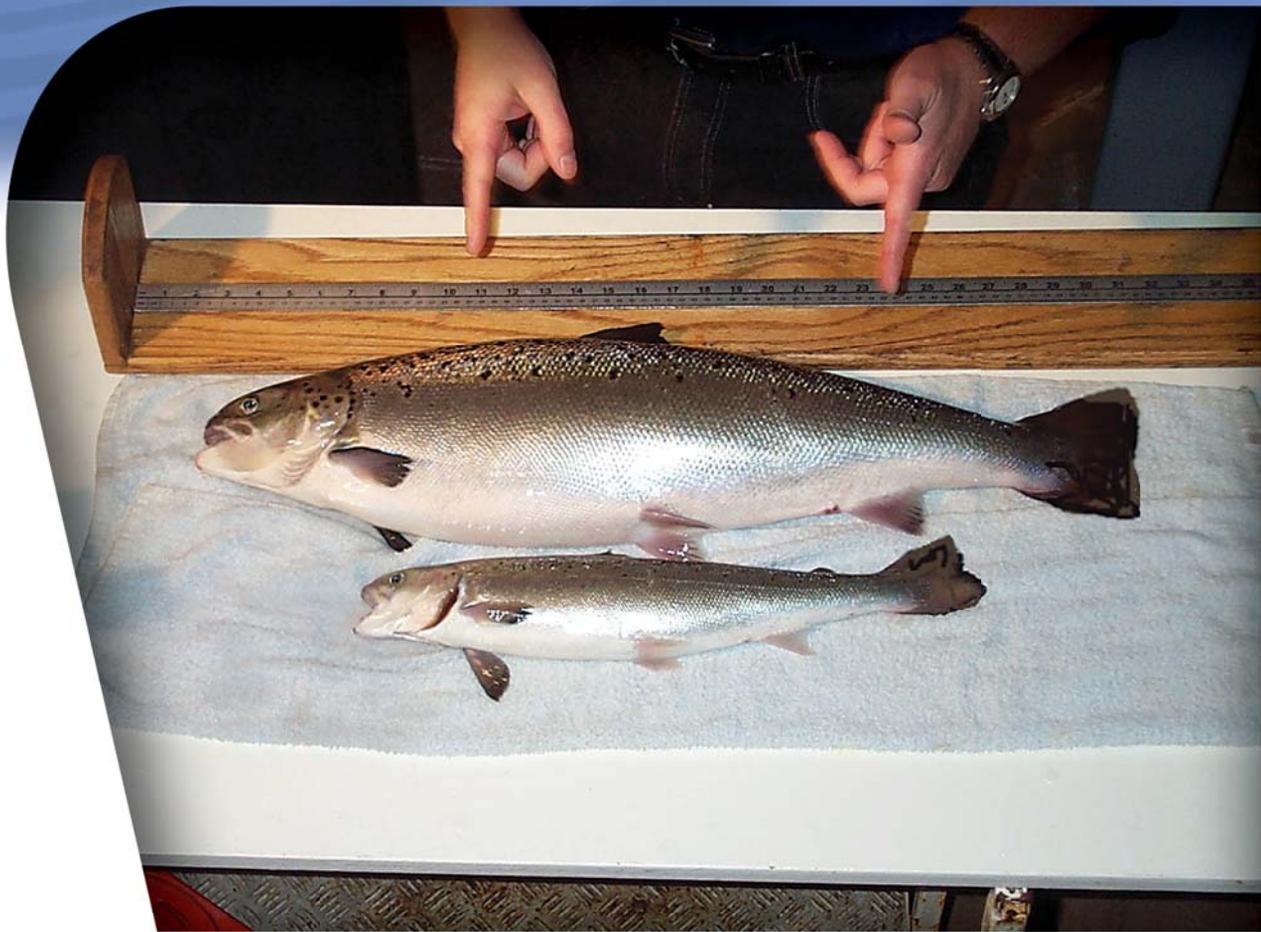
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### Meiosis is the basis of sexual reproduction.

- 6.1 Meiosis
- 6.2 Sexual Reproduction
- 6.3 Human Reproductive Systems
- 6.4 Studying Genetic Changes



# Getting Started



## internet connect

Many crop plants have been genetically modified to change or enhance certain characteristics. Find out which products have been approved by Health Canada for human consumption. Begin your search at [www.discoveringscience9.ca](http://www.discoveringscience9.ca).

**A**re you ready to include genetically modified animals as part of your diet? Genetically engineered plants have been available in supermarkets since the mid-1990s, but Health Canada has yet to approve cloned or genetically modified meat for human consumption. For scientists who have already produced genetically engineered fish and pork products, this approval cannot come soon enough.

All organisms are born with a set of characteristics, or traits, that determine how they look, function, and behave. Reproduction allows these traits to be passed on from an organism to its offspring. In genetic engineering and selective breeding, the set of characteristics that is passed on to an organism's offspring is manipulated to produce desirable characteristics in the offspring. Cloning, on the other hand, produces an exact copy of an organism's entire set of characteristics in its offspring.

In 1989, scientists at Memorial University in St. John's, NL genetically engineered Atlantic salmon by inserting a growth hormone gene from Chinook salmon into fertilized Atlantic salmon eggs. This resulted in salmon that grow faster and process food more efficiently than non-transgenic farmed Atlantic salmon. AquaBounty Technologies was created to commercialize the innovative AquAdvantage Salmon, and is the only company approved to grow these salmon for the market. Some people are concerned that these salmon could pose a danger to native ecosystems by escaping. Environment and Climate Change Canada, Fisheries and Oceans Canada and U.S. Food and Drug Administration have determined the risk of escape to be extremely low.

The Enviropig, produced at the University of Guelph in Ontario, is engineered with ecosystems in mind. It is designed not to grow faster or taste better, but rather to have less of an environmental impact than traditional pigs. Pig manure is usually rich in undigested plant phosphorus, which seeps through the soil and into lakes and streams, where it can upset the balance of an ecosystem and cause harm to fish. The Enviropig is better able to break down phosphorus, so its waste is less harmful to freshwater ecosystems. Consumers, however, are reluctant to eat meat from organisms that have been genetically modified.



The Enviropig releases up to 60 percent less phosphorus in its waste than conventional pigs.

## Designing Your Supper

## Find Out ACTIVITY

Genetic engineering allows scientists to manipulate the characteristics that an organism passes on to its offspring during reproduction. The technology is available to select for desirable traits in food organisms ranging from crop plants to livestock. In this activity, you will evaluate whether genetic engineering technology should be used for this purpose.

### What to Do

1. Working in groups of three or four, choose an organism that is used as a food product. For example, you might choose bananas, peanuts, salmon, or tomatoes.
2. Make a list of five modifications you could make to this organism that would improve it in some way. Consider features that affect product quality, environmental impact, ease of production, and cost of production.

3. For each modification, list possible positive and negative outcomes. Organize your thoughts in a chart like the one below.
4. As a group, come to an agreement about whether the benefits outweigh the drawbacks for each of the modifications.

### What Did You Find Out?

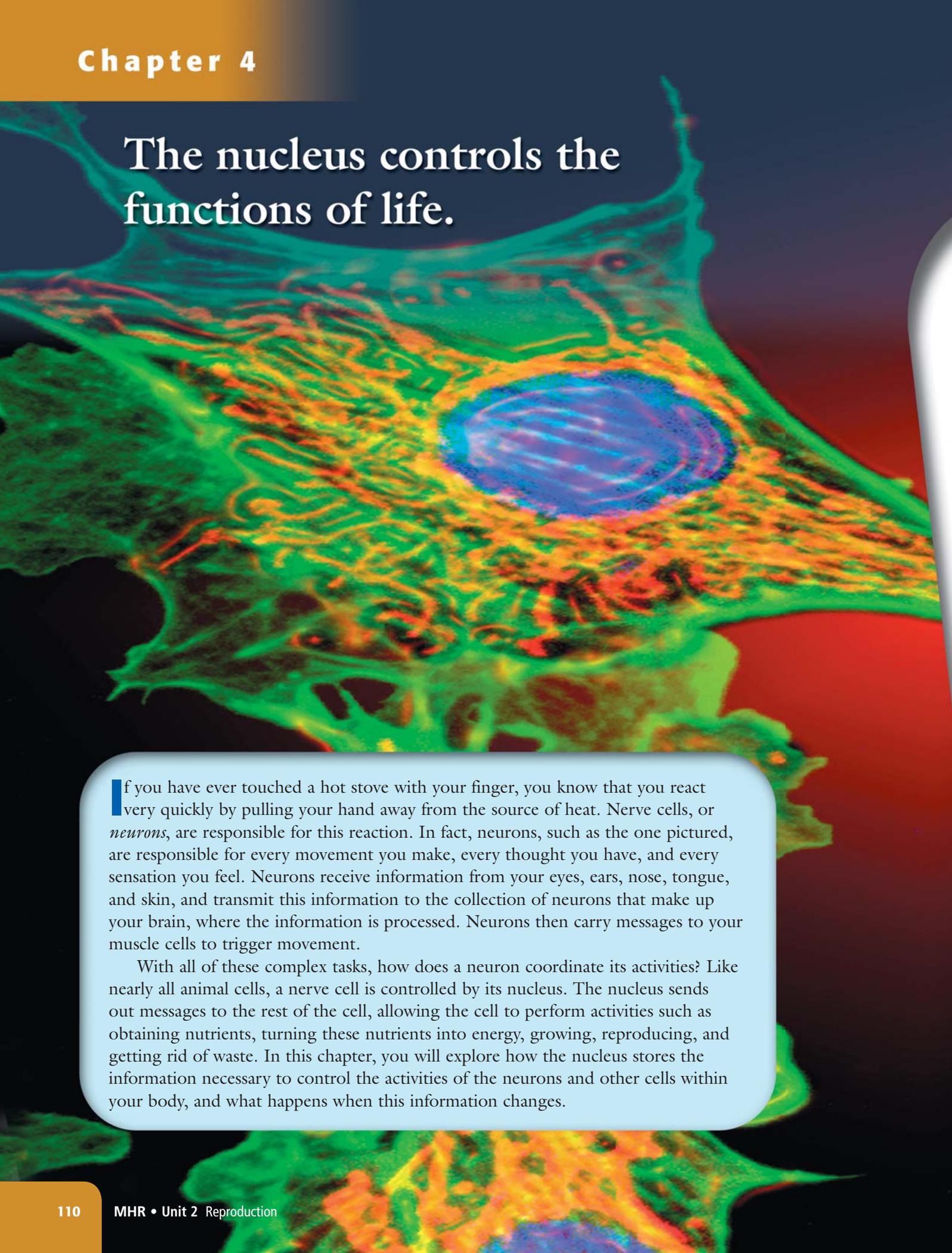
1. Did your group have difficulty coming to a consensus? Explain.
2. Was your decision the same for all of the modifications? (That is, if you felt that the benefits outweighed the costs, was this feeling true for all of the modifications, or did it change on a case-by-case basis?)

Organism

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| Modifications | Positive Outcome | Negative Outcome | Do the benefits outweigh the drawbacks? |
|---------------|------------------|------------------|---|
| 1.            |                  |                  |   |
| 2.            |                  |                  |   |
| 3.            |                  |                  |   |
| 4.            |                  |                  |   |

# The nucleus controls the functions of life.



If you have ever touched a hot stove with your finger, you know that you react very quickly by pulling your hand away from the source of heat. Nerve cells, or *neurons*, are responsible for this reaction. In fact, neurons, such as the one pictured, are responsible for every movement you make, every thought you have, and every sensation you feel. Neurons receive information from your eyes, ears, nose, tongue, and skin, and transmit this information to the collection of neurons that make up your brain, where the information is processed. Neurons then carry messages to your muscle cells to trigger movement.

With all of these complex tasks, how does a neuron coordinate its activities? Like nearly all animal cells, a nerve cell is controlled by its nucleus. The nucleus sends out messages to the rest of the cell, allowing the cell to perform activities such as obtaining nutrients, turning these nutrients into energy, growing, reproducing, and getting rid of waste. In this chapter, you will explore how the nucleus stores the information necessary to control the activities of the neurons and other cells within your body, and what happens when this information changes.

## What You Will Learn

In this chapter, you will

- **describe** the role of the nucleus in controlling cell function, growth, and division
- **distinguish** between genes and chromosomes
- **describe** factors that may lead to changes in a cell's genetic information
- **demonstrate** an understanding of how mutations occur

## Why It Is Important

Understanding the nature of genes and how the nucleus controls cell activities provides us with knowledge about how cells function and how mutations occur. As scientists learn more about the nucleus, they will continue to develop technologies such as gene therapy to correct mutations and treat people with genetic disorders.

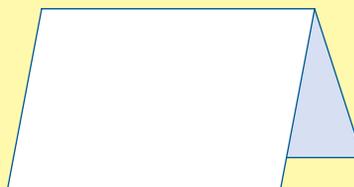
## Skills You Will Use

In this chapter, you will

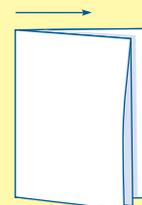
- **classify** gene mutations
- **communicate** your understanding of how the nucleus controls a cell's activities
- **describe** examples of technologies that have developed from an understanding of cellular function.

Make the following Foldable and use it to take notes on what you learn in Chapter 4.

- STEP 1** **Fold** a sheet of letter-sized copy paper in half horizontally.

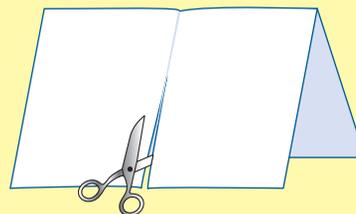


- STEP 2** **Fold** the sheet in half again vertically.

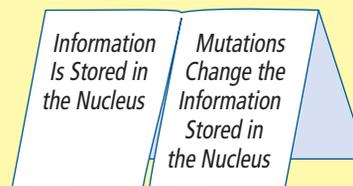


- STEP 3** **Unfold** the paper vertically and leave it folded horizontally.

**Cut** the front tab in half vertically along the vertical fold.



- STEP 4** **Label** the tabs with the following titles: "Information Is Stored in the Nucleus" and "Mutations Change the Information Stored in the Nucleus."



**Organize** Under each tab, summarize information from the chapter using brief points or rough sketches to illustrate the concepts presented.

## 4.1 The Function of the Nucleus within the Cell

The nucleus controls the functions of a living cell. Chromosomes within the nucleus are composed of deoxyribonucleic acid, or DNA. DNA carries the master set of instructions for cell function. Genes are small segments of DNA. Genes contain the information to produce proteins that carry out a cell's activities.

### Key Terms

chromosome  
DNA  
gene  
heredity  
nucleus  
trait

The colour of your eyes, the pitch of your voice, and the size of your nose are all traits that are passed on to you by your biological parents. A **trait** is a particular feature that can vary in size or form from individual to individual within a species (Figure 4.1A). The pattern of traits that an organism demonstrates depends on the information that it inherits from its parents (Figure 4.1B). **Heredity** is the process through which patterns of traits are passed on from an individual to its offspring.



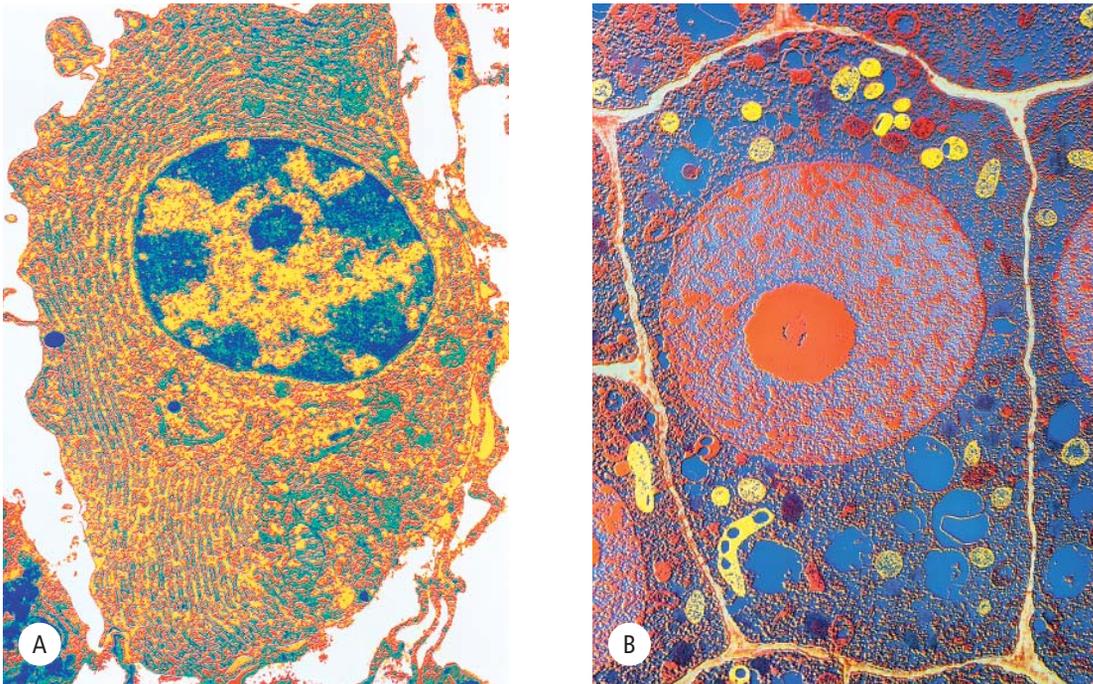
**Figure 4.1A** Each of these basset hounds exhibits slightly different traits, such as ear shape and coat colour and markings.



**Figure 4.1B** The passage of traits from a parent to a child results in family resemblance.

## The Nucleus is Responsible for Heredity

You have already learned that all organisms are made up of one or more cells. You may also recall that both plant and animal cells consist of organelles, such as the cell membrane, cytoplasm, mitochondria, and nucleus. Each of these organelles performs certain functions within the cell. The **nucleus** is the organelle that is responsible for heredity and for controlling the functions of the cell (Figure 4.2). The information contained in the nucleus instructs your cells to produce or import all of the materials they need to survive. This information also determines whether your eyes will be green, blue, hazel, or brown and whether your nose will resemble your mother's or your father's.



**Figure 4.2** The nucleus is a large central organelle found in both animal cells (A) and plant cells (B).

### Reading Check

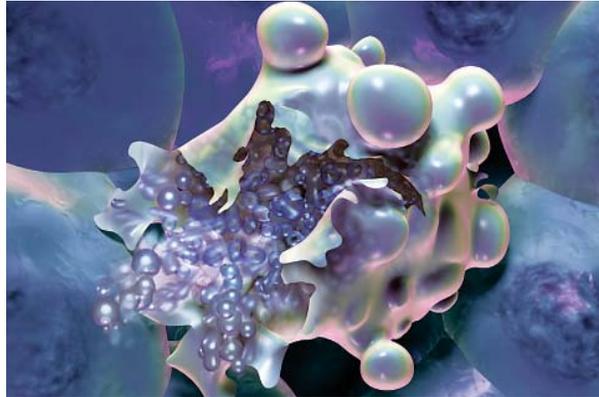
1. Give an example of a trait.
2. What is heredity?
3. Where is hereditary information stored?

## Did You Know?

If the nucleus controls all of the functions within a cell, then it might seem reasonable to expect that every cell in your body contains a nucleus. However, this is not the case. Mature red blood cells do not have nuclei. How do these cells develop, grow, and function without a master set of instructions? When red blood cells form, they do contain nuclei. The nucleus gives the cell the information it needs to develop into an adult red blood cell, but then the nucleus disintegrates to make more space for the cell to carry oxygen.

## The Nucleus: Control Centre of the Cell

Studying the picture on this page and looking up at your teacher require the activity of different cells in the retinas of your eyes. Every cell in your body has a specific function, yet every body cell contains the same cell parts and organelles. So how do retina cells become retina cells and not bone cells? The answer lies in the nucleus. The nucleus contains the master set of instructions that determines what each cell will become, how it will function, when it will grow and divide, and when it will die (Figure 4.3).



**Figure 4.3** A cell in the process of dying. Cell death is important to an organism's growth and life cycle. For example, during your development as an embryo, your fingers started to form when the nuclei in the cells between your fingers instructed these cells to die.

### 4-1A The Nucleus as a Black Box

### Find Out ACTIVITY

Scientists use the term “black box” to describe something in which the inner workings cannot be seen.

A cell's nucleus is a black box, because the activities within the nucleus are not always visible or understood.

In this activity, you will consider the limitations and challenges that a scientist must face when exploring the unknown. Using two different tools, you will investigate the inside of a sealed box. Then, from the information you gain, you will make inferences about the contents of the box.

#### Materials

- 1 black box
- 1 cotton ball
- 1 drinking straw
- clear adhesive tape
- 1 wooden skewer

#### What to Do

1. Your teacher will give you a closed box with a hole in the top. Do not open the box or look through the hole.
2. Attach the cotton ball to the drinking straw with adhesive tape.
3. Put the straw probe through the hole in the box. By feeling around with the straw, determine what is in the box. Draw a sketch of your observations in your notebook.
4. Repeat step 3 with the wooden skewer, using the sharp end. (Do not attach a cotton ball.) Draw a new sketch and add any new information.

#### What Did You Find Out?

1. List as many inferences as you can about the contents of the box.
2. What difficulties did you encounter when you probed the inside of the black box?
3. Explain how you were able to overcome these difficulties.
4. Compare the information you gained using the straw probe with the information you gained using the wooden skewer.

## DNA carries the master set of instructions for cell function

The instructions in the nucleus are carried in long, two-stranded molecules called **deoxyribonucleic acid**, or **DNA**. The DNA molecule looks like a twisted ladder (Figure 4.4). The two strands, or sides, of the DNA ladder wrap around each other in a spiral shape that scientists call a double helix. The word “helix” comes from a Greek word meaning to wrap.

The sides of the DNA ladder are made of sugar and phosphate. The steps of the ladder are made of four nitrogen bases, which are represented by the letters A (adenine), G (guanine), C (cytosine), and T (thymine). Figure 4.5 shows the structure of the DNA molecule.



Figure 4.4 A model of the DNA molecule

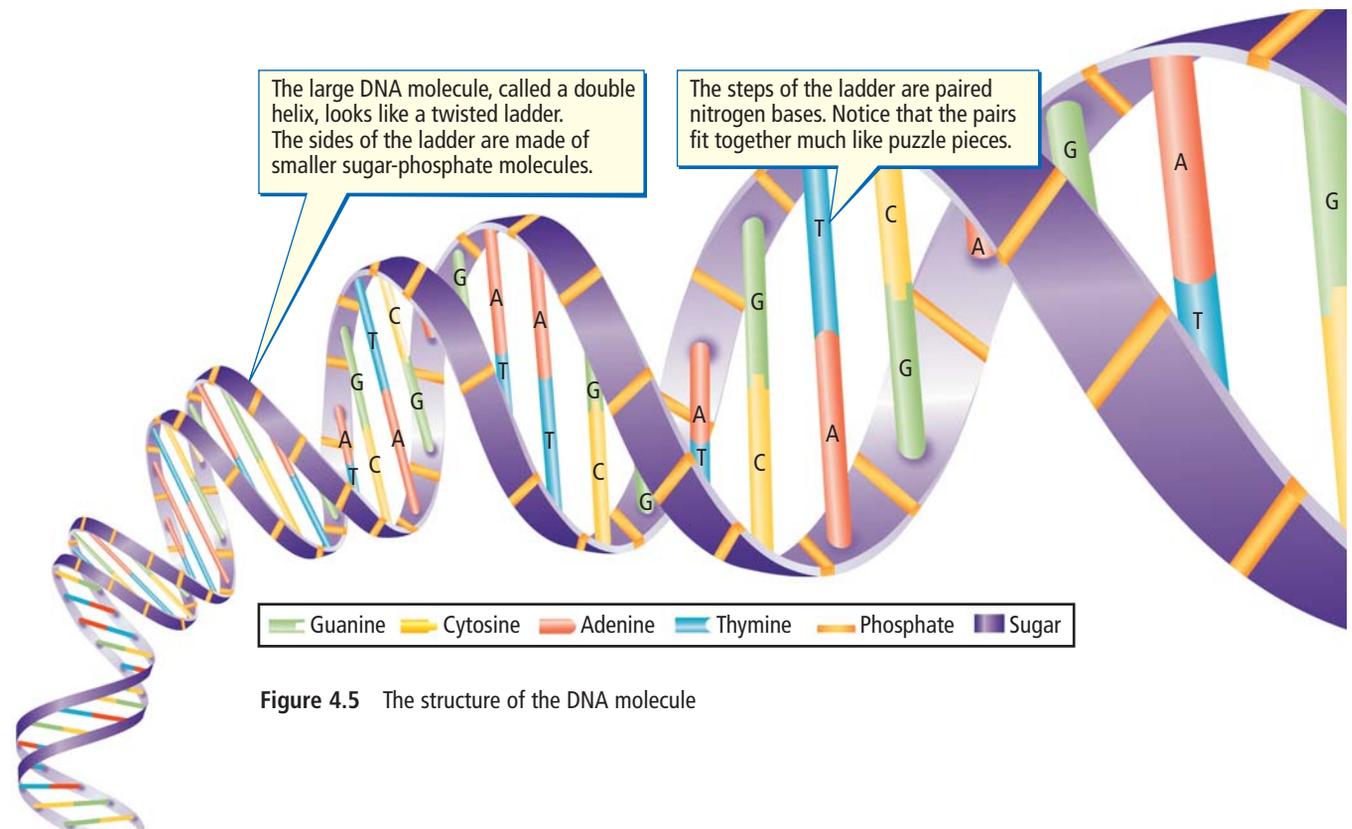


Figure 4.5 The structure of the DNA molecule

## The arrangement of bases in DNA directs all cell activities

Everything that occurs within a cell is the result of how the bases on the DNA molecule are arranged. This arrangement is known as the DNA message. As you can see in Figure 4.5, bases in a DNA molecule always join in a specific way:

- A always joins with T.
- G always joins with C.

However, the order and number of these bases can vary greatly within the DNA molecule. In humans, a single DNA molecule can be several million base pairs in length.

### How is DNA stored?

Most of the time, DNA exists in the nucleus in a loosely coiled form (Figure 4.6). When a cell is growing, the DNA is uncoiled and aids in the manufacture of proteins. Proteins are essential materials required for the cell to carry out the activities necessary for its survival.

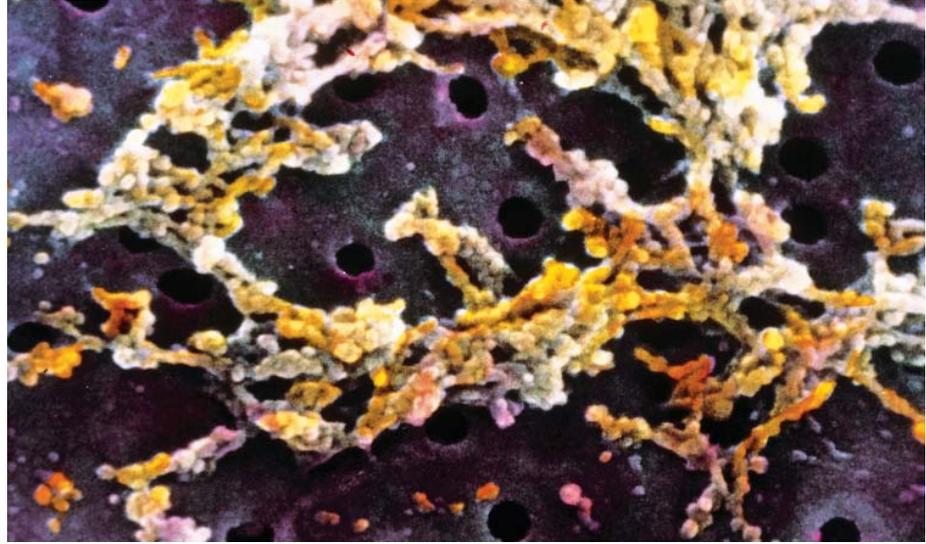


Figure 4.6 DNA is visible in the nucleus in its partially coiled form.

When a cell is ready to divide, each strand of loosely coiled DNA folds up further into a very compact, X-shaped structure called a **chromosome**. Figure 4.7 shows the relationship between DNA and chromosomes.

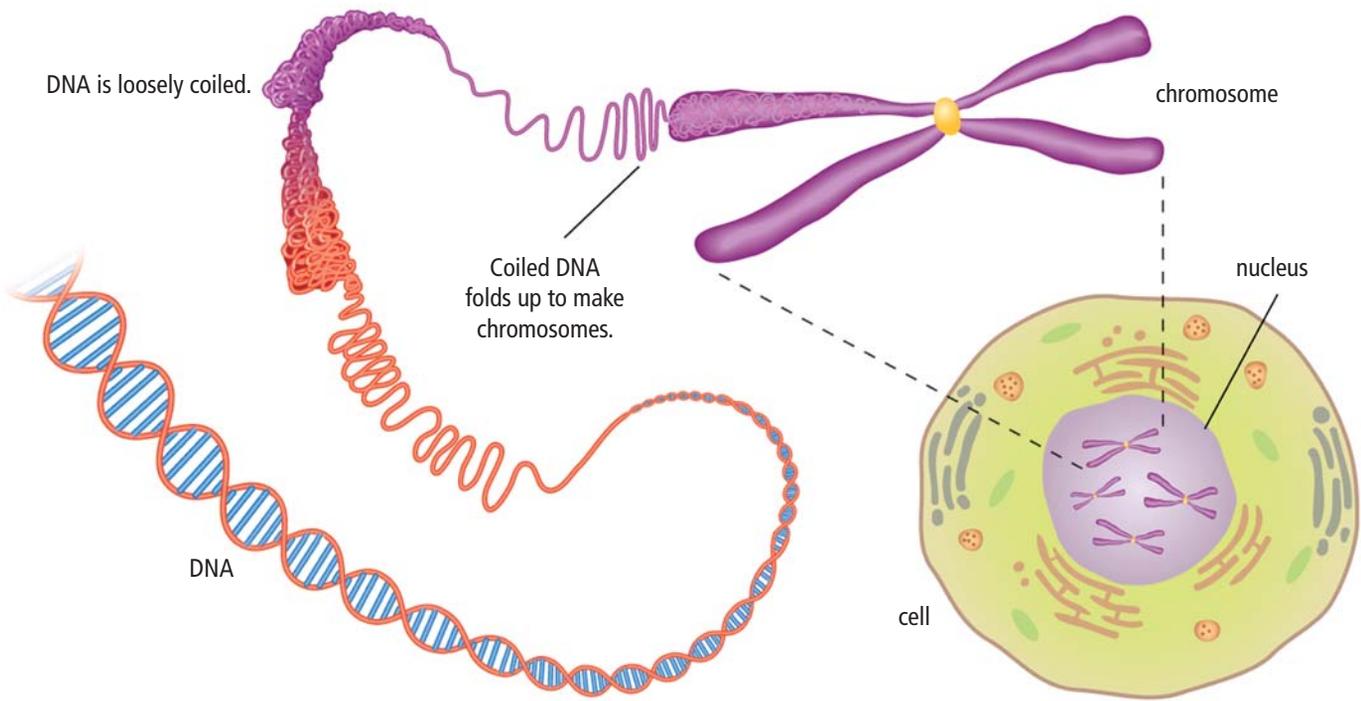
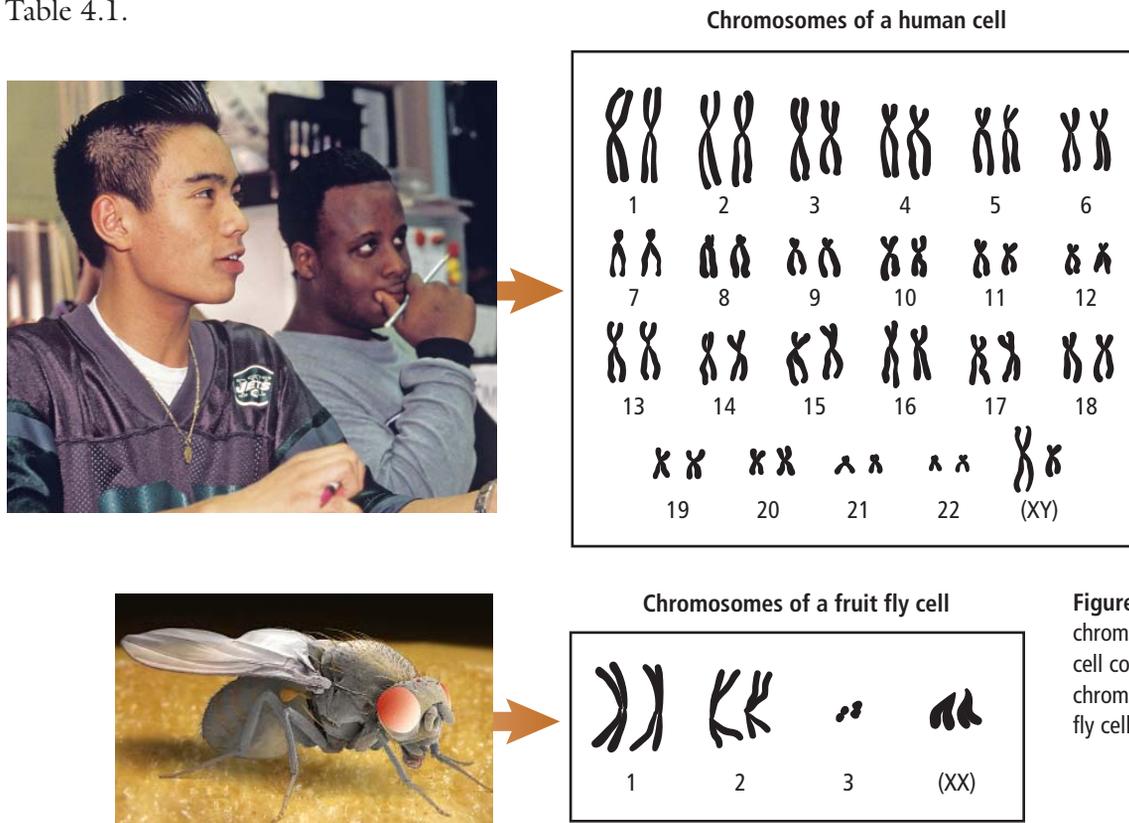


Figure 4.7 In preparation for cell division, DNA is folded into chromosomes.

## Every organism has a characteristic number of chromosomes

Chromosomes within the nucleus are found in pairs. Most human cells have 46 chromosomes arranged in 23 pairs, including one pair of chromosomes that help determine gender (Figure 4.8). In males, the 23rd pair of chromosomes is the XY pair. In females, it is the XX pair. Every living thing has a characteristic number of chromosomes, as shown in Table 4.1.



**Figure 4.8** The chromosomes of a human cell compared with the chromosomes of a fruit fly cell

**Table 4.1** Comparison of Chromosome Number in Various Organisms

| Organism  | Chromosome Number | Organism  | Chromosome Number |
|---|-------------------|---|-------------------|
|  | 46                |  | 20                |
|  | 60                |  | 80                |
|  | 78                |  | 44                |

### Did You Know?

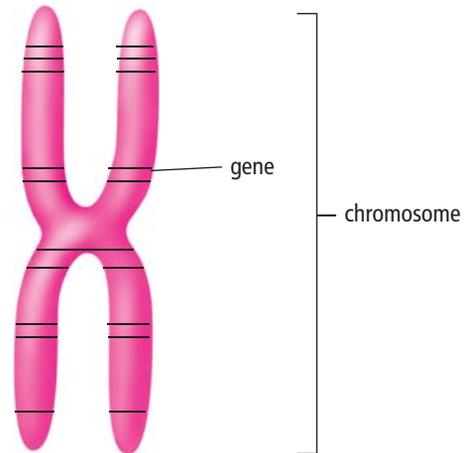
A genome is all the genetic information stored within the chromosomes of a living cell. The Human Genome Project, completed in 2003, found that human cells contain about 3 billion base pairs that carry the information to produce between 25 000 and 35 000 genes. (This estimate has since been reduced to between 20 000 and 25 000 protein-coding genes.)

## Genes are found on chromosomes

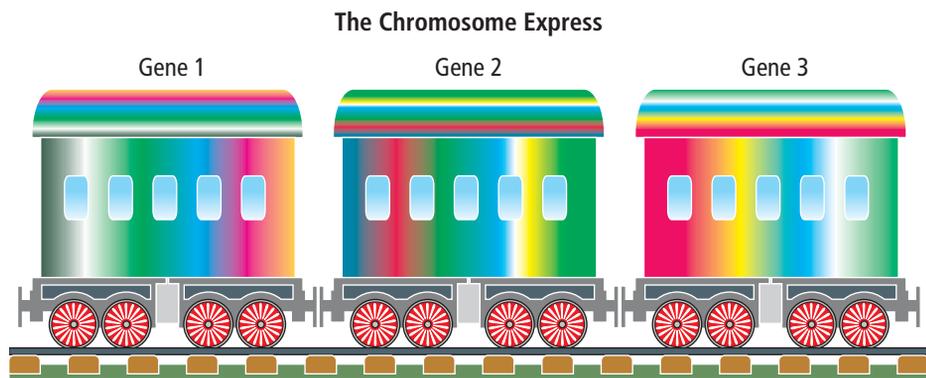
**Genes** are small segments of DNA located at specific places on a chromosome (Figure 4.9). Genes store the information needed to produce 90 000 to 100 000 different proteins used in the cells of your body.

The arrangement of bases in a gene will usually be used to produce a specific protein. Genes can vary in length from hundreds to thousands of bases. Every chromosome carries thousands of genes and therefore contains the information to make thousands of different proteins.

The words “gene” and “chromosome” are sometimes used to refer to the same thing, when, in fact, they have very different meanings. A gene is a section of DNA that contains the coded information to produce a particular trait. A chromosome, on the other hand, contains the coded information for thousands of traits. Chromosomes are made up of genes. If you think of each chromosome as a train, each car of the train represents an individual gene (Figure 4.10).



**Figure 4.9** Genes are located on chromosomes and contain the information to produce a protein.



**Figure 4.10** The image of a train can help you understand the difference between genes and chromosomes.

## Proteins determine what body cells will become and how they will function

Each of your body cells has the same amount of genetic information stored within its 46 chromosomes. However, only specific genes are “read” in each cell to produce specific proteins. By making specific proteins, a cell becomes specialized to carry out a particular function. That is why the cells in your retina are different from the cells in your toenails. Specialized cells come together to form tissues (such as your retinas), and tissues come together to form organs (such as your eyes).

Therefore, proteins needed to make your muscles work are made only in your muscle cells. Proteins needed to help you read this page are made only in the cells of your eyes (Figure 4.11).

Thousands of different, specialized proteins called enzymes speed up the hundreds of chemical reactions that occur within each cell. For example, digestive enzymes work in chemical reactions to break food down into nutrient molecules that provide energy for the cell.

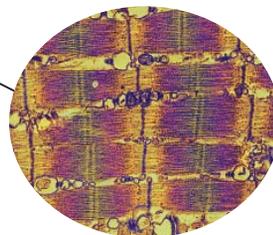
Some proteins act as chemical messengers called hormones. For example, growth hormone functions to prepare a cell for cell division by ensuring the cell has enough nutrients to divide.



Cells in the retina of the eye produce proteins so this skier can see.



Cells in the stomach produce proteins so this skier can digest food.



Cells in the muscles of this skier's legs produce proteins so she can ski.

**Figure 4.11** Although every cell in your body contains the same genes, only certain genes will be read to produce specific proteins, as shown in the three examples on the right.

### Reading Check

1. How many chromosomes does a human body cell contain?
2. (a) What are genes?  
(b) Where are they located?
3. Your retina cells and muscle cells contain identical DNA. How are these cells able to function differently?

## Glowing Genes

The process that makes fireflies glow brightly in the summer night to attract mates has also shone light on the field of biotechnology. Fireflies produce a protein enzyme, called luciferase [lu-SIF-uh-raze], which aids in a light-producing chemical reaction. When living things produce light, the process is called bioluminescence.

Bioengineers have identified the small piece of DNA, called a gene, in the firefly that contains the information to make luciferase. They are able to remove the gene from the firefly, make copies of the gene in the laboratory, and put the gene into another organism. The process where genes are taken from one organism and inserted into the DNA of another organism is called recombinant DNA.

Taking genes from one plant or animal and transferring them into another plant or animal creates a transgenic organism. A transgenic plant or animal has

DNA in its nucleus from another plant or animal. Bioengineers have created transgenic organisms that glow in the dark. Glow-in-the-dark pigs, mice, and monkeys help scientists trace activities going on within a cell. The brighter the light shining from a cell tagged with luciferase, the more activity within the cell.

In the future, scientists may use the process of transferring genes from one organism to another to speed up the testing of new drug and gene treatments. For example, if light shines from the cell that receives the glow-in-the-dark gene, scientists will know that the correctly functioning gene has entered the cell successfully. Gene transfer procedures may be used to treat diseases such as diabetes, AIDS, and cancer. Glowing genes may help identify and treat cells injured by heart attacks or nerve-damaging diseases such as Alzheimer's and Parkinson's. Tumour cells that light up will tell surgeons exactly where to cut to remove a tumour.

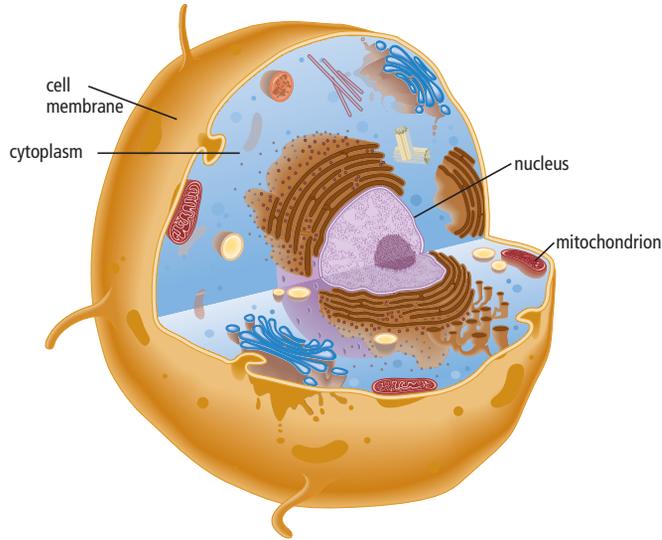
The glowing genes of the firefly hold much more potential for medical discoveries in the future, since they are inexpensive and allow scientists to quickly trace cell activity. Unlike other chemicals used to monitor activity in cells, luciferase is non-radioactive, so it will not harm the organism it is transferred into.



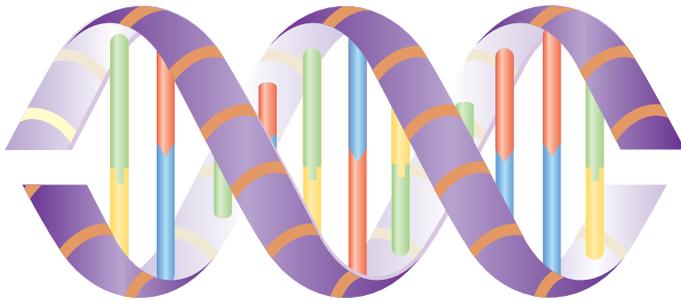
# Check Your Understanding

## Checking Concepts

1. In the diagram of the cell shown below, where would hereditary information be stored?



2. Why is the nucleus sometimes called “the control centre of the cell”?
3. How does a gene differ from a chromosome?
4. Draw a simple diagram of a chromosome and indicate the location of a gene.
5. Explain the function of genes in a cell.
6. What role does the molecule shown below play in heredity?



## Understanding Key Ideas

7. Is family resemblance related to the nucleus? Explain your answer.
8. Explain how the nucleus controls the functions of life.
9. What makes a skin cell different from a nerve cell?
10. The protein hemoglobin, which carries oxygen in red blood cells, is not made in a skin cell. Explain why.
11. Predict what might happen to a cell if a required protein was not made in that cell.
12. Sometimes DNA is loosely coiled, while at other times it is stored in the form of chromosomes. Why do you think this is the case?
13. If you think of DNA as a chemical alphabet, how could you describe genes and chromosomes?

### *Pause and Reflect*

DNA is sometimes referred to as the code of life. Explain why you think scientists have used these words to describe DNA.

## 4.2 Mutation

A gene mutation is a change in the order of the A, G, C, and T bases in a gene. Gene mutations can be positive, negative, or neutral. Mutagens are substances or factors that can cause mutations in DNA. One form of gene therapy is the replacement of a faulty gene with a healthy copy of the gene.

### Key Terms

gene mutation  
mutagen

### Did You Know?

DNA extracted from 43 000-year-old woolly mammoth bones from Siberia suggests that these elephant-like animals may have come in more than the brown variety displayed in museums. By examining genes for hair colour, scientists now conclude that woolly mammoths with blond, red, and black hair also existed.

### Suggested Activity

Find Out Activity 4-2A on page 123



**Figure 4.12** This white kermode bear is an example of a mutation in the gene for coat colour.

The white kermode bear is found in the coastal rainforests of western Canada (Figure 4.12). Known as the Spirit Bear, or Moksgm'ol, the white kermode bear is an important animal in the traditional culture of the Tsimshian First Nation. The Spirit Bear is the result of a gene mutation. A **gene mutation** is a change in the specific order of the A, G, C, and T bases that make up a particular gene. One of the bases may be left out of the sequence, an extra base may be added, or one base may be substituted for another. In the case of the white kermode bear, there is a mutation in a single base in the gene for coat colour found in 1 out of every 10 black kermode bears. Both parents must have this altered gene in order to produce a white bear, and the bear must receive both copies of the gene. To protect the population of Spirit Bears, the population of black bears must also be protected since black bears can carry the mutated gene that produces the white bears.

The Spirit Bear is white because of a gene mutation. The sequence of DNA in a gene is interpreted in groups of three bases. In the kermode bear, the sequence of bases for white coat colour is different from the sequence of bases for black coat colour. Since the DNA sequence is interpreted in groups of three bases, a substitution, loss, or addition of a base will change the meaning of a DNA message. In this activity, you will learn how these three types of gene mutations affect the protein made in a cell.

### What to Do

- To explain gene mutations, scientists sometimes compare DNA sequences to the letters in a sentence. Study the information in the table below. It shows what happens when a letter is substituted into or lost from a sentence and compares these results to what happens in a gene when a base is substituted or lost.

- Copy the following DNA sequence into your notebook. Separate the sequence into groups of three bases.

CATGCCTGACGTCTGATGCCA

- Use the information from the table to help you identify whether each of the following is an example of a substitution, a loss, or an addition of a base. For each example, label where the base mutation occurred on the DNA sequence you copied into your notebook.

- CATGCCTGACCTCTGATGCCA
- CATGCCTGACGTCTGAGCCAA
- CATGCCTGACGTCTGATGGCCA

### What Did You Find Out?

- What types of gene mutations may be the least damaging for a cell? Explain why.
- What types of gene mutations may be the most damaging for a cell? Explain why.

| Example of Sentence Mutations                       | Comparison to Gene Mutations   |
|---|--|
| Themanranforthebus<br>anddidnotgethisdog.           | The original sentence without spaces is like the sequence of bases in a gene.  |
| The man ran for the bus<br>and did not get his dog. | The sentence is read in groups of three letters and makes sense.<br>The DNA sequence is read in groups of three bases and makes the correct protein.   |
| Tee man ran for the bus<br>and did not get his dog. | When only one letter is substituted for another, the sentence is still understandable.<br>When only one base is substituted for another, the gene may still make the correct protein.  |
| Thm anr anf ort heb usa<br>nnd idn otg eth isd og.  | The loss of the letter "e" in the word "the" makes this into a nonsense sentence when the sentence is regrouped into three-letter words.<br>The loss of a base in the DNA sequence of a gene will result in a mutation where an entirely different protein will be made that is not useful for the cell.<br>The addition of a letter in the sentence would also make this example into a nonsense sentence.<br>Similarly, the addition of a base in the DNA sequence in a gene will result in an entirely different protein that is not useful for the cell. |

## Did You Know?

Our genes are estimated to represent only 3 percent of the DNA in our chromosomes. The function of the other 97 percent is under investigation and has been termed “junk DNA” by some scientists. Research has shown that the more complex the organism, the more junk DNA is present. Just as some people keep junk in case they might have a use for it in the future, some scientists think that junk DNA may function as gene backup or provide protection against gene mutation. Research continues in this area.

## The Effects of Mutations

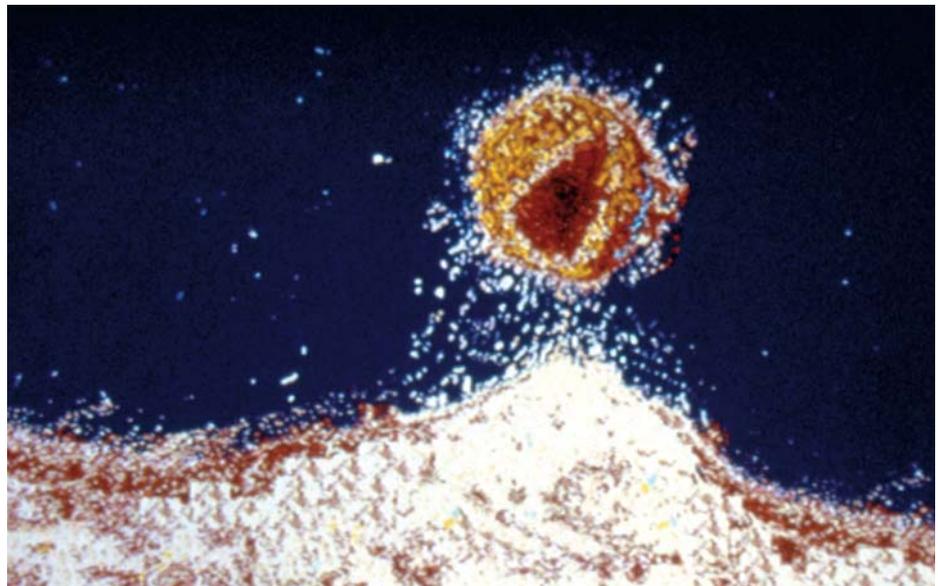
A gene mutation results when the specific order of the A, G, C, and T bases that make up a particular gene changes. A mutation can occur any time in the life of a cell. Types of gene mutations include the following:

- deletion (one base is missing)
- addition (an extra base is added)
- substitution (one base is substituted for another)

Errors in the sequence of DNA bases may produce proteins that could be beneficial to an organism and therefore to the survival of its species. These types of mutation are known as positive mutations. Harmful mutations, known as negative mutations, can cause a species to become extinct. Most often, however, errors in the base sequence of DNA appear to have no effect on the organism. These types of mutations are called neutral mutations.

### Positive mutation

Millions of people worldwide are infected with HIV (human immunodeficiency virus). As a result, millions of people will develop AIDS (acquired immune deficiency syndrome). Figure 4.13 shows an active HIV particle. There is still no known cure or vaccine (a substance that provides immunity against infection) for HIV and AIDS. However, a few individuals have been found to be resistant to the virus and, therefore, to AIDS. These individuals carry a mutated gene that produces the instructions for a protein that prevents HIV from infecting the person. This type of mutation, which benefits an individual, is an example of a positive mutation.



**Figure 4.13** An HIV particle bursts out of a kind of white blood cell known as a T cell. HIV infects T cells and causes them to produce more HIV cells. In individuals who carry the positive mutation, the HIV particle is prevented from infecting a T cell.

Some plants have developed resistance to bacteria and fungal infections. Positive mutations create proteins that are beneficial to the plant and protect them from disease-causing invaders, or pathogens (Figure 4.14).



**Figure 4.14** The plant on the far left has a positive mutation, which protects it from getting the disease affecting the other three plants.

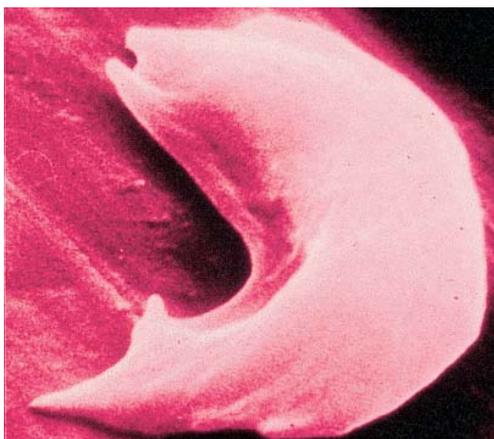
 **internet connect**

There are other types of mutations that can alter the information in DNA. Find out more by going to [www.discoveringscience9.ca](http://www.discoveringscience9.ca).

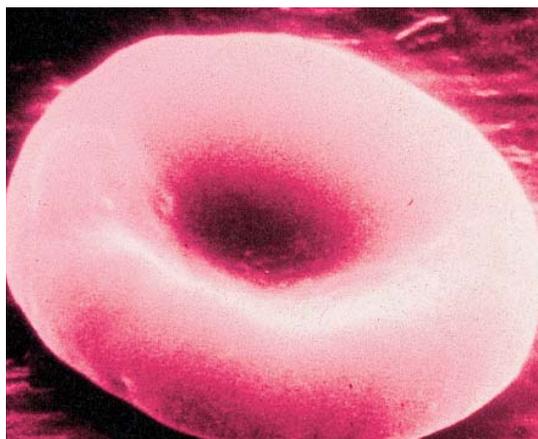
### Negative mutation

Small changes in the sequence of bases can also cause a harmful or negative mutation. Negative mutations reduce the probability that organisms with the mutation will produce offspring or survive in their environment. The substitution of the base A for the base T in only one position on the gene causes the protein hemoglobin to take on a different shape. This differently shaped hemoglobin molecule causes sickle cell anemia (Figure 4.15). Abnormally shaped molecules cannot carry oxygen efficiently. They also block blood flow, causing pain and often organ damage since blood carrying nutrients cannot reach organs such as the lungs, liver, and kidneys.

(A)



(B)



**Figure 4.15** People who carry the sickle cell gene have red blood cells that are C-shaped (A). Normal red blood cells are disc-shaped (B).

## internet connect

Canadian scientists helped discover the cystic fibrosis gene. Find out more about how they found the location of the gene and what the work of molecular geneticists involves by going to [www.discoveringscience9.ca](http://www.discoveringscience9.ca)

Cystic fibrosis is another genetic disease caused by a mutation. In fact, the disease can be caused by more than 1300 different mutations in one gene. BREATHE (Basic Research and Therapy) is a Canadian research program set up to expand current knowledge about the gene responsible for cystic fibrosis. In cystic fibrosis, mucus builds up because the protein that normally functions to transport chloride ions into and out of the cell is not made correctly. Since the protein malfunctions, chloride ion levels build up, affecting the thickness of mucus in the lungs, causing respiratory problems, and making breathing difficult. Daily antibiotics and physical therapy are necessary to prevent lung infection (Figure 4.16). Mucus build-up also prevents pancreatic juice from flowing to the small intestine.



**Figure 4.16** To control the effects of mucus build-up in the lungs, cystic fibrosis patients receive physiotherapy to clear air passages.

### Neutral mutation

Just as the substitution of one letter in a sentence may not change its meaning, the substitution of one base for another in the DNA sequence of a gene may not change an organism. The same protein will be made and may still function normally. For example, if a mutation occurs in a gene for brown coat colour in mice, the gene may still produce the same brown pigment. The change caused by the mutation does not increase or decrease the survival rate of the organism. This type of mutation, which does not affect the organism, is called a neutral mutation. The gene mutation that results in the white coat colour of the Spirit Bear is considered to be a neutral mutation. This mutation neither increases nor decreases the survival rate of the Spirit Bear.

## Mutagens

**Mutagens** are substances or factors that can cause mutations in DNA. Just as a computer virus can disrupt the instructions in computer software, a biological virus can disrupt the instructions stored in genes. By attaching to the DNA, biological viruses can cause genes to be misread or copied incorrectly. Cigarette smoke, radiation from X rays or UV rays, pollutants such as mercury, and even household chemicals are examples of environmental mutagens that can cause mutations.

When DNA becomes damaged, the proteins in a cell will not be made correctly. In Chapter 5, you will learn how these non-functioning proteins may upset the life cycle of a cell and cause cancer.

### Reading Check

1. Why is the Spirit Bear white?
2. What is a gene mutation?
3. How do viruses cause mutations?
4. Describe two sources of mutagens.
5. List three examples of environmental mutagens.

### Did You Know?

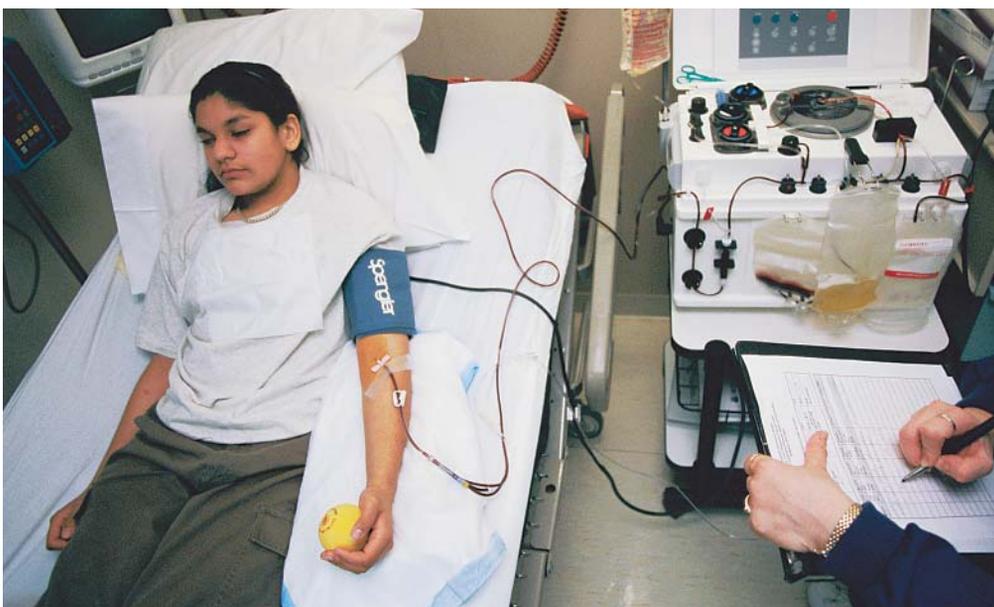
Some mutagens occur in nature, while others are the result of human activity. For example, UV rays, gamma rays, and X rays are naturally occurring sources of radiation that can cause damage to DNA. Humans are responsible for producing cigarettes, cleaning products, and industrial wastes that may contain chemical mutagens.

Learn how dangerous cigarettes are and how some tobacco companies lure young people into tobacco addiction.

Begin your search at [www.discoveringscience9.ca](http://www.discoveringscience9.ca).

## Correcting Mutations

You have read how some gene mutations can cause disease. At the moment, the most effective way to treat these conditions is to use drugs or surgery. Researchers are testing new techniques called gene therapy to treat mutated genes. In one form of gene therapy, researchers replace a mutated gene with a healthy copy of the gene. Because the technique is risky, gene therapy is currently being tested on diseases without known cures (Figure 4.17).

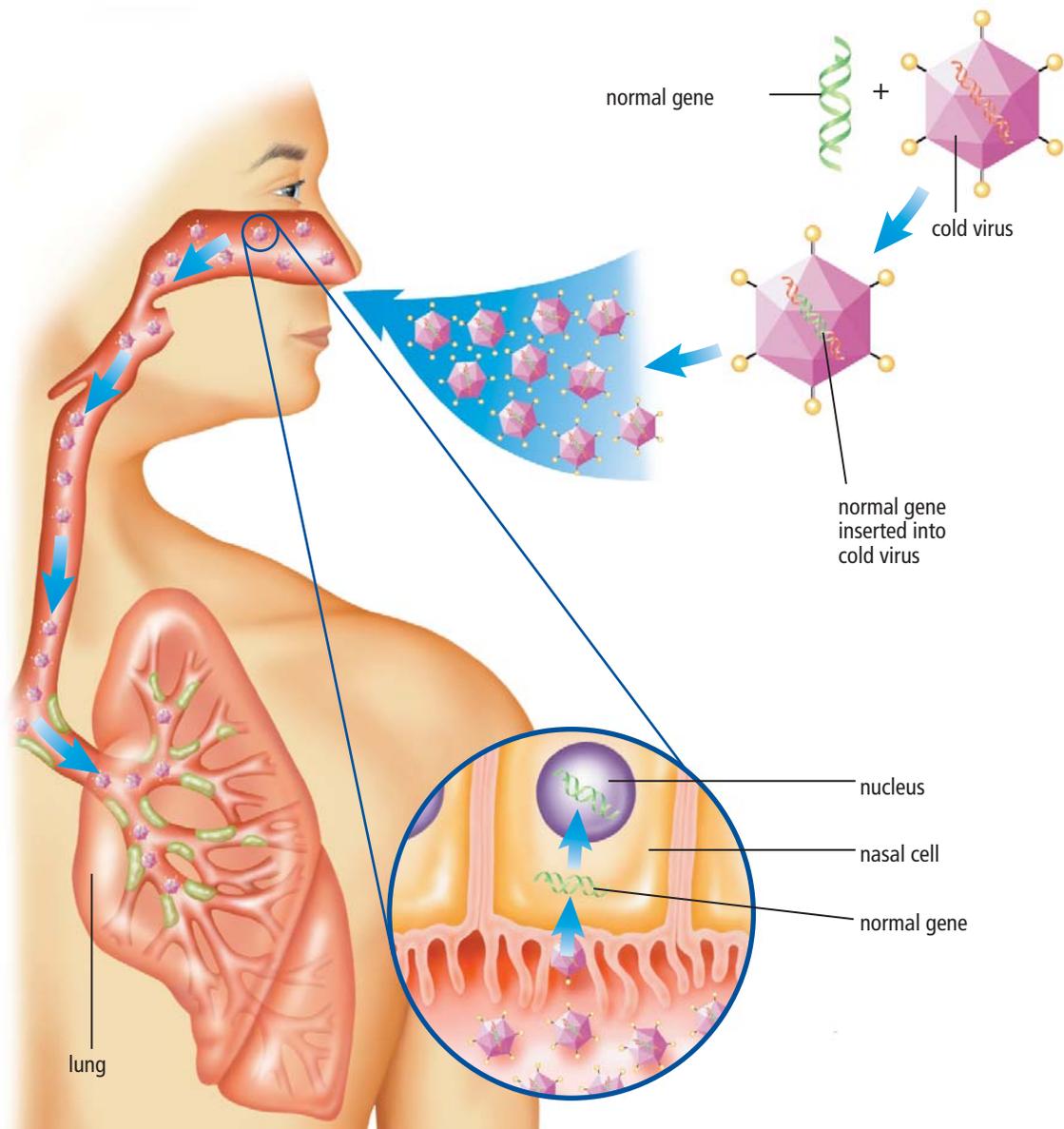


**Figure 4.17** Ashanti DeSilva was the first person ever to receive gene therapy. Researchers hoped that gene therapy would cure Ashanti's rare immune disorder by reprogramming her defective immune system cells to produce healthy cells. This did not occur, and Ashanti still receives gene therapy to help control her disorder.

**Suggested Activity**

Think About It 4-2B on page 129

Gene therapy is not simple and is a highly experimental procedure. A geneticist requires the skill of an archer shooting an arrow toward the target. The target is cells with faulty, mutated genes. The arrow used is often an inactive virus, which carries the healthy gene to the diseased cells (Figure 4.18). However, the healthy gene will be useful only if the geneticist can shoot the arrow—the inactive virus—into the nuclei of millions of target cells. Then the patients’ cells must be able to activate or “switch on” these healthy genes so that the cells produce the healthy protein. To switch on the gene, the healthy gene must first become attached to a chromosome within the nuclei of a patient’s cells. To be successful, the information on the healthy gene must be read in order for the healthy protein to be made. The healthy gene must also be able to make the correct amount of the protein.



**Figure 4.18** An inactive cold virus is taken into the body and delivers the “healthy gene” to cells affected by cystic fibrosis.

Gene therapy can affect an individual's immune system, and some patients have not responded well to treatment. In 2006, however, researchers announced that gene therapy used to treat melanoma (cancer that begins in the skin) had been shown to boost a patient's immune system. By genetically altering white blood cells called T-lymphocytes, researchers have been able to increase the ability of a cancer patient's immune system to fight advanced melanoma.

## 4-2B Considering Gene Therapy

## Think About It

Although gene therapy has been successful in some cases, patients do face certain risks when participating in research trials. Some people are also concerned that gene therapy will be used for gene enhancement. For example, making athletes' muscles stronger would give them an unfair advantage over other athletes. In this activity, you will consider the pros and cons of gene therapy.

### What to Do

1. Examine your beliefs regarding gene therapy by completing the survey that follows. Assign a rating to each statement using the following scale.
  - 1 is strongly disagree
  - 2 is somewhat disagree
  - 3 is neutral
  - 4 is somewhat agree
  - 5 is strongly agree
2. Provide a reason for the rating you assigned to each statement.
  - (a) Everyone should have equal access to gene therapy treatment no matter what the cost if gene therapy is known to cure a disease.
  - (b) Because of the risks, gene therapy testing should not be conducted on humans until gene therapy is proven 100 percent safe and effective.
  - (c) If gene therapy is proven safe, then it is acceptable to use gene therapy to enhance a person's genetically inherited characteristics, provided the individual pays all expenses for the treatment.
  - (d) Gene therapy should be used only as a last resort to treat critically ill patients.

### What Did You Find Out?

1. Summarize in a paragraph what you have learned about your beliefs regarding gene therapy.

# Science Watch

## Banana Factories for Vaccines

A mother brings her baby to a clinic in Mozambique, Africa, but knows there is little the doctors can do for her child. Her baby's skin and eyes have turned yellow. The baby will not eat and continues to cry. Why? Because the baby contracted the virus hepatitis B from his mother during childbirth. The baby would survive if he had been vaccinated. A vaccination would have made him immune to the disease. However, even though the hepatitis B vaccine has been available for 25 years, it is still not readily available in developing countries. About 20 percent of infants in developing countries go without vaccinations, resulting in more than 2 million deaths per year.

Currently, vaccines cost \$50 to \$100 per child. There is controversy about the chemicals used to preserve vaccines, and many vaccines must be refrigerated. In addition, setting up clinics to provide vaccinations is often challenging in developing countries. But biotechnologists may have the answer. They are currently developing a painless, inexpensive protection against hepatitis B. Instead of receiving a needle with the vaccine, children will be given a banana to eat.

Biotechnologists are injecting genes from the hepatitis B virus into young banana trees. Only genes that make the protein coat of the virus are transferred to the banana. As the banana tree grows, the cells of the banana begin to make this protein. When a person eats one of these bananas, the protein is absorbed through the intestine into the blood. Since the protein is a foreign substance, the person's white blood cells will produce antibodies against this protein invader. This response is similar to what happens when a vaccine is injected. If a hepatitis B virus enters the body, the antibodies made in response to the protein will attack the incoming virus.

There are several advantages to using bananas to produce vaccines. Bananas can be eaten raw. Other staple foods such as rice and potatoes require cooking, which could destroy the protein. Bananas can also be easily mashed for infants. It would cost only a couple

of cents for each vaccine since one banana-producing plant could grow more than 45 kg of bananas. Whereas other vaccines require refrigeration and medical staff to administer them, banana vaccines do not require either. In addition, developing countries could grow their own bioengineered bananas. However, since banana crops take 3 years to grow, scientists predict an edible vaccine is at least 10 years away.



## Questions

1. Calculate how many children die each day in the developing world because they were not vaccinated.
2. Explain how a person who ate a bioengineered banana would be protected from a virus.
3. What are the advantages of using bananas for vaccine production?

# Check Your Understanding

## Checking Concepts

1. How can a gene mutation create a change in an organism?
2. Why is it important to protect the black kermode bear in order to protect the Spirit Bear?
3. What is a mutagen?
4. Describe an example of a mutation caused by a naturally occurring mutagen.
5. Describe an example of a mutation caused by a human activity.
6. What can happen when a mutated gene makes a protein that doesn't function properly? Provide an example.

## Understanding Key Ideas

7. Mutations to DNA cause changes in organisms. Explain why this might be important to the survival of a species.
8. Predict whether a mutation in a human skin cell that results in cancer can be passed on to an offspring. Explain.
9. A field of wheat is killed by a disease. Among the dead stalks, three remain alive and healthy.
  - (a) Explain how the genes of these three plants may have helped them survive.
  - (b) How might a farmer use the surviving plants to prevent that disease from killing the crop in future years?

10. Why is it important to be protected with a lead shield when you are getting dental X rays?
11. Can you protect yourself from all mutagens? Why or why not?
12. Why should you always use proper precautions when handling chemicals?
13. Why can suntanning be dangerous for a skin cell?
14. Are all mutagens harmful? Use an example to support your answer.
15. Electromagnetic radiation is used in many medical tests. These include mammograms, which screen for breast cancer, and CT scans, which are often used to diagnose lung, liver, or pancreatic cancer. Do you think the benefits of these tests outweigh the risks? Explain.

## *Pause and Reflect*

Since cigarettes contain mutagens, to what degree should smoking be restricted? Should the use of tobacco be banned completely? State your opinion, then write a paragraph to support it.

### Prepare Your Own Summary

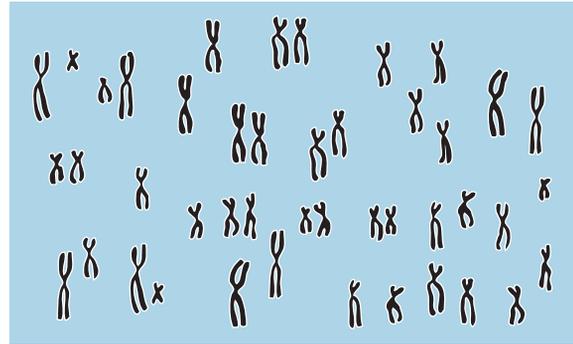
In this chapter, you investigated the function of the nucleus within the cell and the function of genes in creating proteins. Create your own summary of the key ideas from this chapter. You may include graphic organizers or illustrations with your notes. (See Science Skill 8 for help with using graphic organizers.) Use the following headings to organize your notes:

1. The Nucleus: Control Centre of the Cell
2. Genes
3. Mutations

### Checking Concepts

1. In what way is the nucleus like a black box?
2. Explain why DNA is required in every cell.
3. Give three examples of human traits.
4. How is the nucleus related to heredity?
5. How many chromosomes are in a human skin cell?
6. What are the functions of proteins in cells?
7. What is the difference between DNA and chromosomes? Draw a simple diagram to support your answer.
8. Explain why the correct sequence of DNA bases is important for the production of proteins.
9. Describe the results of two known gene mutations.
10. What causes gene mutations?
11. How can we protect ourselves from mutagens in our environment? Use examples to support your ideas.

12. A cell was broken open and its chromosomes were observed under a microscope. A sketch of the chromosomes is shown below. Was the cell taken from a human? Explain.



### Understanding Key Ideas

13. How are traits related to genes?
14. You are reading a magazine article that talks about “the unit of heredity.” Is it likely referring to a DNA molecule, a gene, a chromosome, or a genome? Explain.
15. Compare the amount of DNA in a skin cell to the amount of DNA in a muscle cell.
16. How does DNA direct a cell to become a muscle cell or a stomach cell?
17. Do you think the number of chromosomes in an animal cell or a plant cell reflects how advanced the organism is? Why or why not?
18. What could happen if the nucleus of a cell did not perform its function?

19. How can a gene mutation affect the survival of a species?
20. Some occupations pose greater risks of mutation to DNA than others. List five occupations that you think pose a risk of mutation, and explain the reason for your choices.
21. Are all of an organism's characteristics likely controlled by the cell nucleus? Explain.
22. Examine the photographs below. List the source(s) of potential mutagens shown in each photo, and state whether they are naturally occurring or human-made.

(a)



(b)



## *Pause and Reflect*

In previous studies, you have learned that the nucleus is the control centre of the cell. In this chapter, you have seen how the nucleus controls the functions of life. What new information did you learn in this chapter that has added to your knowledge about the nucleus and how it controls cell functions?