

## Key Terms

chromosome  
DNA  
gene  
DNA screening  
transgenic organism  
cloning  
mutation  
mutagen

**chromosome** in a cell nucleus, a thread-like structure made mostly of DNA

## 1.2 Genes: Answers and Questions


As shown in **Figure 1.8**, babies in Canada are normally tested soon after birth for a genetic condition known as PKU, the short form for phenylketonuria [pronounced fen-il-KEE-to-NU-ria]. If uncorrected, PKU can lead to severe brain damage. However, if diagnosed promptly, PKU can be corrected. Treatment includes following a diet very low in natural protein. (People with PKU must obtain their protein in a special liquid form.) Understanding what causes this condition involves knowing what happens deep within the cell—in the nucleus.

### The Nucleus: Control Centre of the Cell

The nucleus contains the master set of instructions that determines what each cell will become, how it will function, and how long it will live before being replaced. These instructions are carried in **chromosomes**. Every plant and animal species has a specific number of chromosomes in the nucleus of each cell. In the cells of most plants and animals, chromosomes come in pairs—one of each pair comes from each parent when an egg and a sperm unite to produce a fertilized egg. Humans, for example, normally have 46 chromosomes in the nucleus of each body cell, 23 from the mother's egg and 23 from the father's sperm. As shown in **Table 1.1**, other species have their own specific number of chromosomes.

**Table 1.1** Comparison of Chromosome Numbers in Various Organisms' Cells

Organism	Chromosome Number (per cell)	Organism	Chromosome Number (per cell)
Human	46	Corn	20
Cow	60	Butterfly	80
Chicken	78	Fruit fly	8



**Figure 1.8** To test a baby for PKU, a blood sample is normally taken from the baby's heel in the first few days after birth, and then again about a week later.

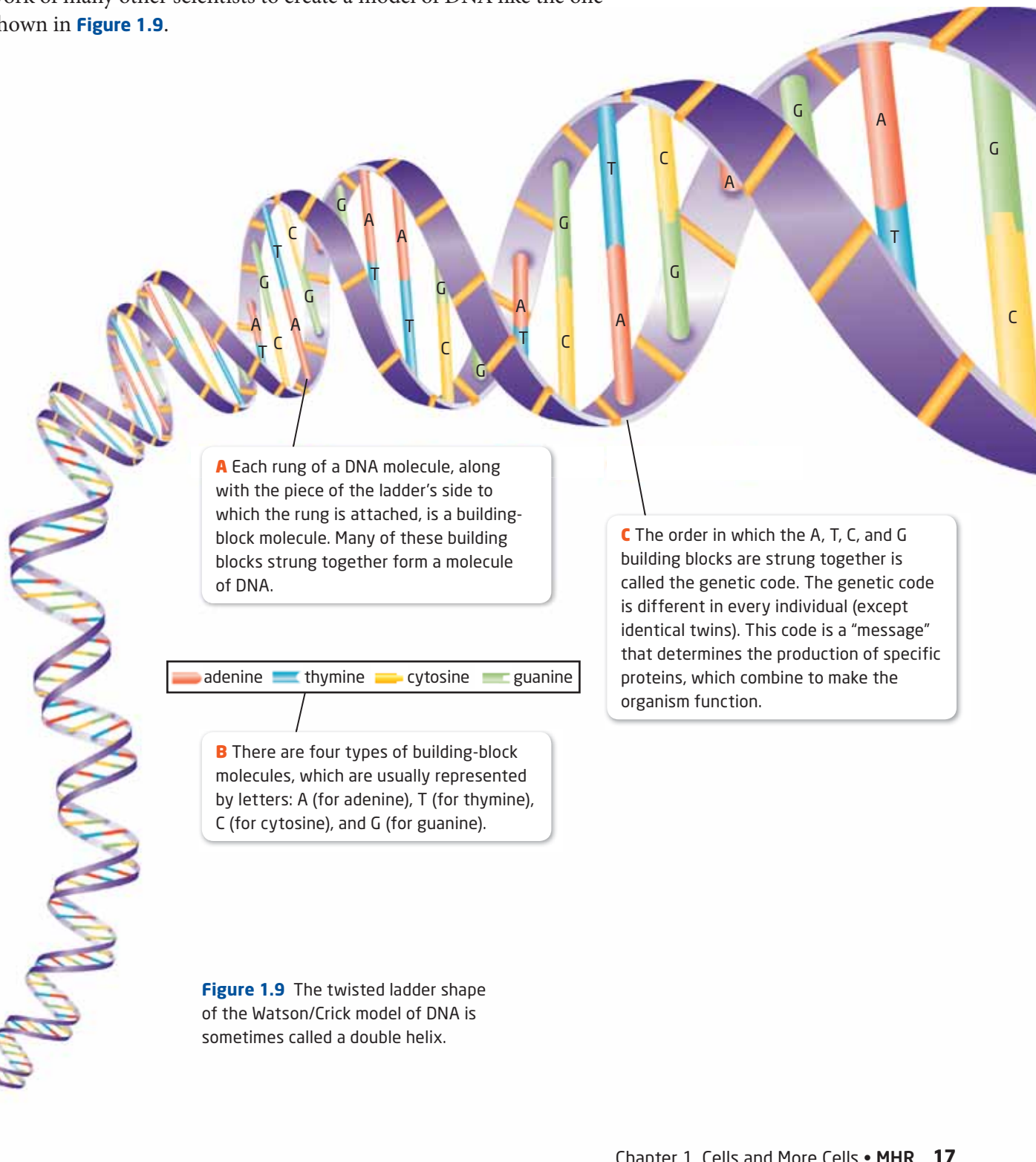
## The DNA Code

Chromosomes are made of a material called deoxyribonucleic acid (**DNA**). In fact, each chromosome consists of a single molecule of DNA, which is divided into segments called **genes**. Genes are located in specific places on the DNA molecule. Most genes provide instructions for making proteins. Thus, genes control the cell's activities, and much of its structure, by controlling what proteins are made and when.

In 1953, scientists James Watson and Francis Crick built upon the work of many other scientists to create a model of DNA like the one shown in **Figure 1.9**.

**DNA** material found in the cell nucleus that contains genetic information

**gene** a segment of DNA that controls protein production

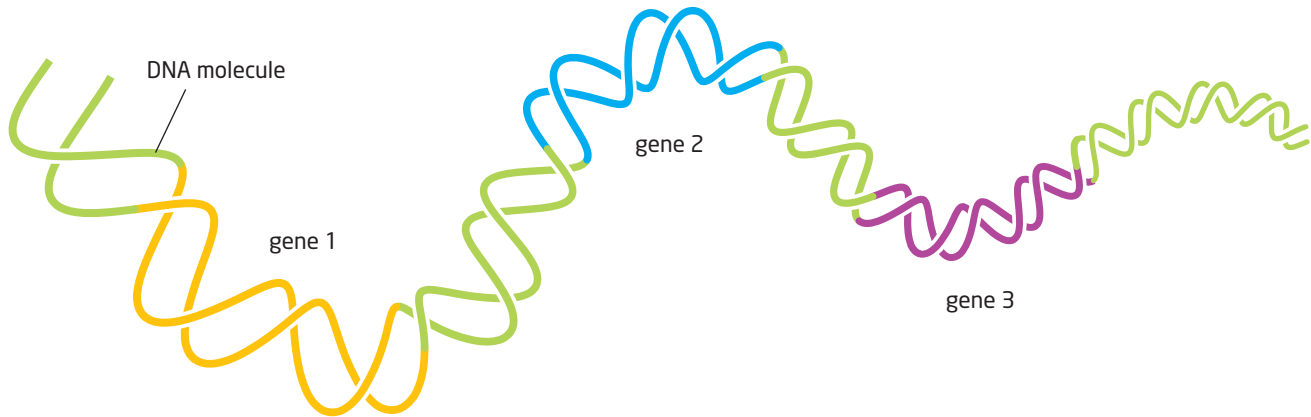


**Figure 1.9** The twisted ladder shape of the Watson/Crick model of DNA is sometimes called a double helix.

## Why Is DNA Important?

As shown in **Figure 1.10**, a gene is a relatively small part of a DNA molecule. Each DNA molecule contains hundreds or thousands of genes. DNA controls many of your features, such as your hair and eye colour, and whether you can digest certain foods, such as milk. Your DNA exerts this control through genes—they determine what kinds of proteins your cells can make, and therefore how your body might function or look. Of course, your lifestyle choices, such as what and how much you eat and how much exercise you get, also play a role in how your body functions and looks.

**Figure 1.10** This diagram shows a model of a section of a DNA molecule with three genes labelled. Genes vary in length, but each is probably thousands of building-block molecules long.



### Sense of **scale**

Our genes are estimated to represent only 3 percent of the DNA in our chromosomes. The function of the other 97 percent is currently unknown.

## Protein Production

As you have just read, the job of genes, and thus of DNA, is to control the manufacture of proteins. Each protein is designed to do a specific job. For example, some proteins help build parts of your body. Others are like couriers, carrying materials short or long distances within your body. Some pick up or transfer signals from one body part to another. Still others, called enzymes, catalyze (speed up) various chemical reactions in your body, such as those that help you digest food. All of these various proteins get their “orders” from DNA.

### Learning Check

1. Write a sentence that shows the relationship among the following terms: DNA, gene, protein.
2. What is the role of genes in the cell?
3. Use **Figure 1.9** to explain how genes vary.
4. How do you think it is possible for a genetic code with only four components (A, C, T, and G) to control the production of thousands of different kinds of proteins? It might help to think of the building blocks as letters of an alphabet, and to remember that each gene is made of thousands of building blocks.

## DNA Screening

On page 16, you learned that babies are commonly tested for a genetic condition called PKU. Testing for the presence of genetic disorders is referred to as genetic screening, or **DNA screening**. Some types of genetic disorders can be observed by looking at a person's chromosomes.

Down syndrome is one of these disorders—it can even be detected in a fetus. Using a technique called amniocentesis, a needle is inserted through a pregnant woman's abdominal wall to withdraw a sample of fluid from the amniotic sac (which surrounds the growing fetus). Cells from the fetus are isolated, and a micrograph of the chromosomes in these cells is taken.

Although this micrograph, called a *karyotype*, cannot show errors in individual genes, it can show if a person has too many or too few chromosomes, or if any are broken. To diagnose Down syndrome, technicians look specifically at chromosome 21. Individuals with Down syndrome, such as the girl shown in **Figure 1.11**, have three of these chromosomes instead of the usual pair (two).

### Testing for PKU

Other kinds of genetic conditions, such as PKU, can be detected by examining a blood sample. In such tests, the presence or absence of specific proteins in the blood can indicate whether a person's genes are functioning normally. If a certain protein is present at normal levels, the gene must be functioning properly. To test for PKU, for example, a baby's blood is examined for an enzyme needed to digest certain kinds of protein in foods. Without this enzyme, a substance called phenylalanine builds up in the baby's blood, with tragic consequences if uncorrected. Each year, one baby in 12 000 is born with PKU.

Testing for PKU is inexpensive compared to the costs of treating PKU if it is not detected early. Other types of genetic screening, however, are more expensive and the benefits may be less clear. For example, having certain genes may make a person *more likely* to get cancer or heart disease, but this does not mean that the person *will* get the disease. The person might lead a healthy lifestyle and be in little danger. Genetic information alone cannot provide *all* the answers.

**Figure 1.11** This girl has Down syndrome, a genetic disorder resulting from having 47 chromosomes instead of 46. This extra chromosome leads to overproduction of certain proteins, which results in some physical and developmental disabilities.

**DNA screening** the process of testing individuals to determine whether they have the gene or genes associated with certain genetic disorders

### Study Toolkit

#### Previewing Text Features

Notice the two heading styles on this page. What do these different styles tell you about the relationship between the information in each section?





## Testing for Huntington Disease

A look at the case for and against genetic testing for Huntington disease reveals some of the ethical issues at stake in DNA screening. Huntington disease is a genetic disorder that affects nerve cells. Symptoms, which normally appear in a person's 40s, include loss of muscular control and brain function. The symptoms worsen for 15 years or so before the disease causes death.

Scientists have identified which gene causes this disease, which helps researchers such as Nancy Wexler, shown in **Figure 1.12**. Individuals have a 50 percent chance of having the gene that causes this disease if one of their parents has it. Someone who has the gene *will*, with certainty, develop the disease. Therefore, finding out if the gene is present in a person's body leads to a definite diagnosis.

### Should Individuals at Risk Get Tested?

There is an ongoing debate about whether individuals at risk of Huntington disease (that is, people who have a parent with the disease) should get tested. Those against testing argue that diagnosing the disease can cause people needless emotional pain—since there is no cure, there is nothing they can do. Moreover, critics argue that testing is too expensive, especially since it will not save lives.

Those in favour of testing argue that having the test results—positive or negative—reduces the stress of uncertainty. In addition, knowing whether they do or do not have the gene might change many of a person's life decisions. If they test positive, for example, some people may choose not to have children (so they do not risk passing on the gene). However, even people who support the tests argue that the results must be kept strictly private so that individuals who test positive are not discriminated against.

**Figure 1.12** Much research on Huntington disease has taken place in San Luis, an isolated village in Venezuela, where the gene is very common. One of the people leading research there is Nancy Wexler, whose mother had the disease. Here she stands in front of a chart she created to track the genes of a group of families from New York City.



## Activity 1-2

### To Test or Not to Test?

Globally, Huntington disease affects about seven in 100 000 people, although some locations have a higher incidence than others. Genetic testing for a disease like Huntington raises many complex ethical issues. What kinds of issues might these be?

#### Procedure

1. Choose the perspective of a stakeholder in the debate about testing for Huntington disease. Suggested roles include the following:
  - You are a 30-year-old who has one parent with the disease. You are wondering whether to be tested.
  - You have just tested positive for the disease. You have a two-year-old child.
  - You are the owner of an insurance company considering a new client's application.
  - You are an employer. Your company invests heavily in employee training and generally counts on people staying with your company for many years or even decades.
  - You are a government official in charge of public health care. You are looking for ways to control costs.
2. Make a list of points to support your opinion using this textbook and your own ideas. Prepare a summary statement of your position.
3. In your role, discuss the questions that follow in a small group.
4. Share your ideas as a class.



In your group, try to reach a consensus on each question.

#### Questions

1. Should employers or insurance companies be allowed to require people to test for Huntington disease, or should taking the test be a matter of personal choice? Explain your answer.
2. Should people be required to reveal a family history of Huntington to a potential employer or insurance company? Or should such a matter be private?
3. Should children with a risk of Huntington disease be allowed to have the test? Why or why not?
4. Should health-care professionals discourage people with a risk of Huntington from having children? Explain your answer.
5. Should individuals pay to be tested, or should the public health-care system pay? How might this decision affect access to the test?
6. Should all stakeholders have an equal voice in decisions about genetic testing for Huntington disease? Explain your answer.

### DNA Screening in Canada

PKU, Down syndrome, and Huntington disease are not the only conditions that can be identified using DNA tests. For example, women can be screened for the presence of certain genes associated with breast cancer. Although only a small proportion of all breast cancers are due to genetic factors, the test could help women with the gene take preventative measures. Other conditions that can be determined with DNA screening include cystic fibrosis and spina bifida.



**Figure 1.13** In 2007, the Food and Drug Administration in the United States approved Kuvan, a drug to treat PKU. For an adult, the annual cost of taking this drug might be as high as U.S. \$200 000.

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## Ethical Issues and Drug Research

Much of the research into treatments for various diseases, including PKU and Huntington disease, is carried out by drug companies. Such research can be very costly and take years, if not decades. Once a company develops a drug that is effective and safe in lab conditions, it applies to carry out a clinical trial on humans. In Canada, approval for drug trials is given by Health Canada, a department of the federal government. The financial risks for a company involved in this type of research are very high, since the drug might not work as well outside the lab. As discussed in **Figure 1.13**, companies may try to recover the costs of research and development by putting a high price on their product.

Critics sometimes suggest that clinical trial results can be biased. Companies are sometimes accused of presenting their trial data as more positive than they really are. The ethical issues get even more troublesome if you think about what might happen if a company discovered an effective, but extremely expensive, cure for cancer or diabetes. Does the company own the cure and therefore have the right to sell or control it in any way it chooses? Or does the company have an ethical obligation to make life-saving cures available to everyone who needs them? These ethical questions are frequently debated by scientists, medical professionals, drug companies, and concerned members of the public.

### Learning Check

5. What is a karyotype?
6. Describe two techniques used to screen a person's DNA.
7. How can having too little or too much of a certain protein cause problems for an organism?
8. Every container of diet soft drink that is sweetened with the chemical aspartame must have on its label a warning that it contains phenylalanine. Why do you think this warning is required?

## Altering Genes: Benefits and Controversies

The genetic code is universal. This means that the same four DNA building-block molecules (A, C, T, and G) produce the code for proteins in *all* types of organisms, including bacteria, plants, and animals. Theoretically, this means that the genetic code in one type of organism could be “read” by any other type of organism. If a particular gene could be transferred between two different types of organisms, one species could then make proteins usually made only by the other species.

In fact, scientists have been combining the DNA from different species for a number of years in a process called genetic engineering. The species whose genes are altered are often called genetically modified organisms (GMOs) or **transgenic organisms**.

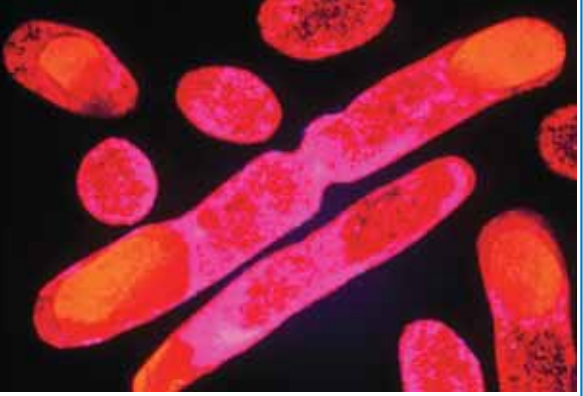


**transgenic organism** an organism whose genetic information has been altered with the insertion of genes from another species

## Transgenic Organisms

**Table 1.2** shows some of the common kinds of transgenic organisms today. Many people see the manipulation of genes as a way to solve various problems. Others, however, worry about the effects of causing rapid change in species that have taken thousands, if not millions, of years to evolve. They feel that we know too little about the long-term consequences. For example, some people ask how GMO plants might affect the organisms (including humans) that eat them. Others worry about the effects of GMO plants on the ecosystem, especially if the GMO plants spread to new areas.

Some people ask whether transplanting organs from transgenic animals, such as the pigs you saw on page 4, is a good idea. They worry about various viruses that are carried in pigs without harming them, but that are not naturally found in humans. If these viruses moved with the transplanted organs, they could cause viral diseases that could spread quickly in the human population.

**Table 1.2** The Uses of Some Genetically Modified Organisms

GMO Organism	Benefit	Example
<p>Bacteria injected with human proteins are used for many medical treatments. Bacteria have a relatively simple genetic structure, so they were the first type of organism to be genetically modified. Here the bacteria <i>E. coli</i>, shown in magenta, has been genetically engineered to produce insulin, which is used to treat diabetes. Orange areas indicate insulin production sites on the bacteria.</p>	<p>Human proteins manufactured by bacteria are less likely to cause allergic reactions or diseases, compared with proteins obtained from other sources, such as animals or dead bodies.</p>	
<p>Many crops have had genes from bacteria or other plants inserted into their chromosomes. Corn, canola, wheat, cotton, and soy are just some of the crops for which farmers can buy GMO seed. Pictured here is a plot used to promote a company's GMO product.</p>	<p>Crops can be modified in this way to resist specific pests, to have a higher nutritional value, or to better withstand drought (lack of water) or cold.</p>	
<p>Some animals are injected with genes that code for a hormone that promotes growth. Here, Canadian scientist Robert Devlin holds up a 16-month-old GMO salmon on the left and a normal salmon on the right.</p>	<p>GMO animals injected with a growth hormone grow faster than non-GMO animals, increasing human food supplies.</p>	



**cloning** the process of creating identical genetic copies of an organism

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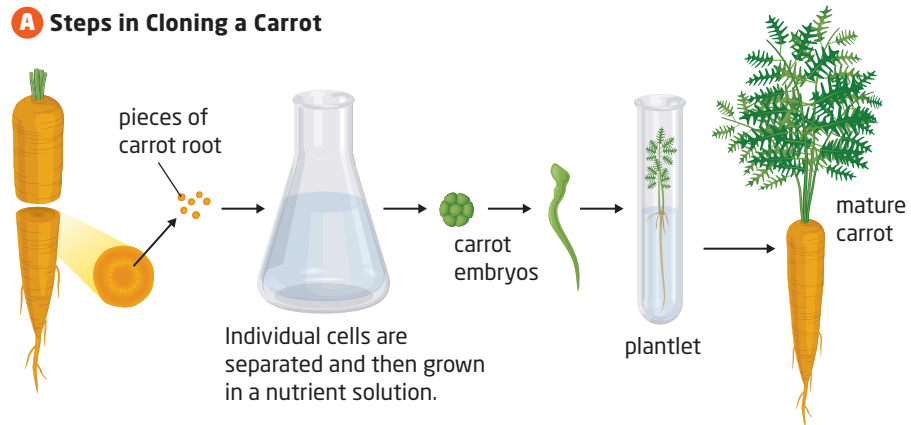


## Cloning

**Cloning** may be another solution to the problem of needing new organs or cells. Cloning is the process of producing identical offspring from genes, cells, or an entire organism.

Cloning has been used for centuries in its simplest form—by gardeners who have taken cuttings from a plant, rooted them, and thereby produced more plants that are exact copies of the parent plant. Then, in 1958, Fredrick Steward was able to grow a plant from a single carrot cell in a laboratory. **Figure 1.14** shows a simplified version of Steward's cloning experiment (**A**) as well as a process used to clone a mammal today (**B**).

### **A** Steps in Cloning a Carrot



**Figure 1.14** In **A**, you can see the cloning process used by Fredrick Steward. In **B**, you can see a process used to clone a mammal, which was first accomplished in 1996.

## STSE Case Study

### Clones in the Kitchen

Why would we clone the animals we use for meat? The purpose of cloning, which is a type of biotechnology, is to produce genetic duplicates (copies) of animals that are considered superior in some way. That way, desirable traits that occurred naturally, such as higher quality meat, are passed along from generation to generation. Cloning animals benefits meat and milk producers because high-quality products mean higher profits.

Also, altering the genetic material from the donor animal before the animal is cloned provides more opportunities to

incorporate benefits. For instance, cattle could be genetically engineered to better resist BSE (mad cow disease). However, animal cloning opponents are concerned about food safety. Few scientific studies have examined cloned meat, and livestock companies have done most of the research. Since these companies want positive results, some people believe their research may be biased.

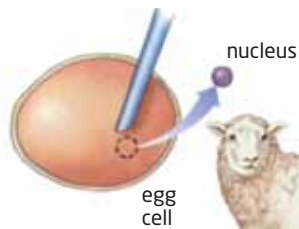
Some studies show that even though cloned animals are genetic copies of the donors, they are not completely identical. For example, meat from some cloned animals has higher levels of fat compared with meat from the original donor animals.



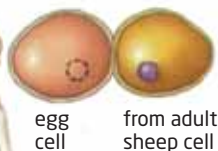
Some meat and milk producers want to use cloned animals as a way to guarantee the quality of their products.

## B Steps in Cloning a Mammal

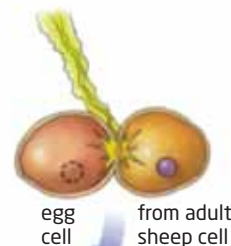
1. Scientists remove the cell nucleus of an egg from a female sheep.



2. A cell is removed from an adult sheep. This cell and the egg cell are placed next to each other in a bath of chemicals.

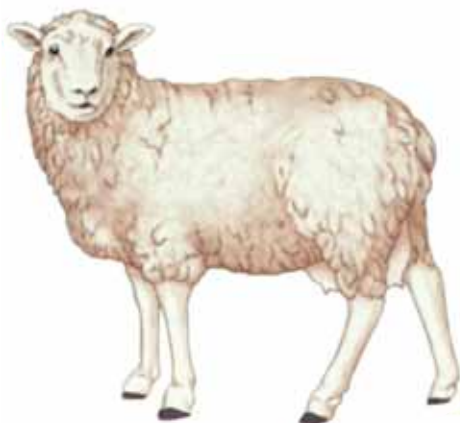
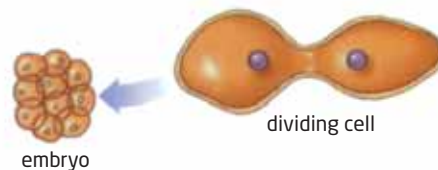


3. A jolt of electricity causes the two cells to fuse.



5. The embryo is then inserted into the uterus of a surrogate mother to complete its development. The resulting lamb is a clone of the sheep that donated the adult cell.

4. The fused cell begins dividing to form an embryo.



Furthermore, genetically *identical* animals are more prone to catch the diseases that animals around them have. That is, if a virus or other micro-organism infects one of these animals, it could easily infect a whole herd. In nature, a herd would include genetically *different* animals and would usually include some animals whose immune systems are better able to fight the disease.

Because we know so little about using cloned animals for food, Canada currently bans the sale of food from cloned animals or their offspring. Where do you stand on this issue?

### Your Turn

1. Survey the other students in your class. Ask them, "If you eat meat, would you consider eating the meat of a cloned animal? Why or why not?" Calculate the percentage of "yes" and "no" answers you receive.
2. Suppose that Health Canada has decided to permit the sale of cloned meat. Identify your position on this issue. Provide three pieces of supporting evidence.
3. In a small group, brainstorm and record the positive and negative characteristics of cloned animals in a two-column table. In a brief report to the class, present your group's evaluation of whether animals should be cloned.



In 1996, a sheep named Dolly became the first mammal created from a cell of an adult sheep rather than from an egg. Dolly died at the age of six. Some people claim her life was shorter than normal because she was a cloned animal, while others argue that her death was completely natural.



### Learning Check

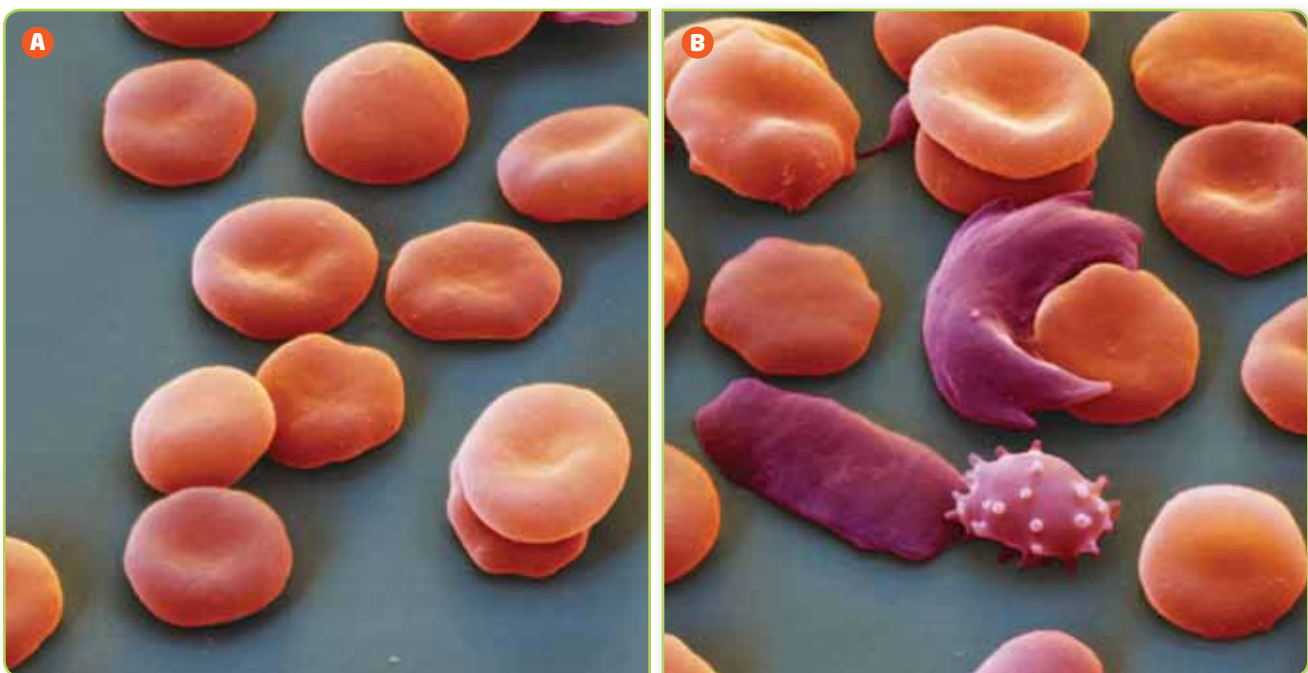
9. Why is the genetic code described as being universal?
10. In what way are organs from certain kinds of transgenic organisms potentially significant to human health?
11. Traditional Aboriginal ethical values stress protection for the rights of future generations, acceptance of human limitations, and a precautionary approach to new science and technology. How might such values affect someone's view on transgenic organisms or cloning?
12. Think about what you have eaten today. Do you think any of it was genetically modified, or contained any genetically modified ingredients? If so, what might those ingredients be? Look back to **Table 1.2** for ideas.

**mutation** a change in the DNA of an organism

**Figure 1.15 A** Normal red blood cells are disk-shaped. Their shape enables them to move easily through blood vessels. In **B**, you can see a C- or sickle-shaped red blood cell from someone who carries the sickle cell gene.

### Mutations

As you have learned, the specific protein that a gene codes for depends on the order of the DNA building blocks in the gene. A change in the usual order of the A, C, T, G building blocks is called a **mutation**. A mutation in a gene can alter the structure of the protein it produces. Such a change can then affect how well the protein does its job. For example, people who have a condition called sickle cell anemia have a mutation in the gene that codes for hemoglobin, a type of protein in red blood cells. As discussed in **Figure 1.15**, the mutation changes the protein in such a way that it is less able to perform its function, which is to help the blood carry oxygen to body cells.



## How Mutations Happen

The sequence of the A, C, T, and G building blocks in a DNA molecule can change for no particular reason. Often, however, mutations are caused by **mutagens**, forces or substances that physically damage DNA. Electromagnetic radiation, including X rays and UV rays (from the Sun) are mutagens. So are many chemicals, including mercury and the tar in cigarettes. As shown in **Figure 1.16**, some mutations occur in only one or a few of an organism's cells. Other mutations, however, affect cells (eggs and sperm) that are passed between generations. In other words, some mutations are inherited.

Not all mutations are harmful. Only part of a DNA molecule contains genes—the rest does not code for proteins. A mutation that occurs on a non-genetic part of a DNA molecule is not harmful, at least as far as scientists know. Even mutations that change proteins are not necessarily harmful. Some mutations, for example, might help an organism adapt to a particular environment, such as the mutations in bacteria that allow them to resist antibiotics. Other mutations may be neutral—neither causing a problem nor helping the organism. Whether the mutation is harmful or not may sometimes depend on an organism's environment. Do you think the mutation shown in **Figure 1.17** is beneficial, harmful, or neutral?

## Could Science Correct Mutations?

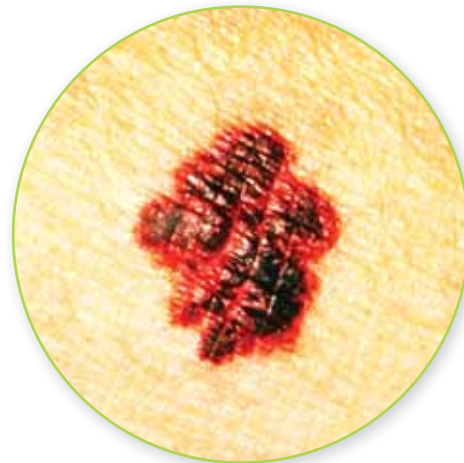
Gene mutations are at least partly to blame for a variety of diseases. Research into gene therapy—correcting faulty genes—is ongoing. Researchers hope that gene therapy will one day be used to treat a variety of inherited diseases.

However, clinical trials on various forms of gene therapy have not yet had much success. People who participate in the experimental treatments often become sicker or die. Some people question whether this kind of research is even worth pursuing. For them, the issue is who decides what disorders should be corrected. For example, researchers have had some success using gene therapy for certain types of inherited blindness. Does the fact that this blindness can be “fixed” imply that these people are “broken”? Many people would disagree and argue that visually impaired people can live full, rich lives. Other questions include the following:

- Who gets access to treatments, and who pays for the treatments?
- Who funds research, and who owns or manages the resulting product or technology?
- How far should society go in using available technologies?

Some people believe these questions should be answered before gene therapy treatments are developed.

**mutagen** a substance or factor that can cause a mutation in DNA



**Figure 1.16** This photograph shows skin cells that have been mutated by UV radiation. This mutation would affect only the cells of this person; the mutation would not be passed on to the person's children.

**Figure 1.17** This albino American alligator has a mutation in the gene that codes for the proteins that produce colour in its body.





## Section 1.2 Review

### Section Summary

- The nucleus of a cell contains chromosomes, which are composed of DNA and divided into segments called genes. Genes control a cell's structure and function by controlling the production of proteins.
- There are many ethical issues related to technological developments in DNA screening, transgenic research, and clinical drug trials. Researchers often make progress faster than society's ability to make decisions on how to use such research.
- Mutations in genes change the structure and function of the proteins in the cells. Many, but not all, genetic mutations are harmful.

### Review Questions

- K/U** 1. Why is the nucleus of a cell so important?
- K/U** 2. Draw a simple diagram of a portion of a DNA molecule, and indicate the location of a gene.
- C** 3. How does DNA screening have both positive and negative implications for society? Create a T-chart to summarize your answer.
- A** 4. How might researchers who work for pharmaceutical companies ensure that their clinical trials meet proper scientific standards?
- C** 5. As of 2007, 23 countries grew genetically modified crops. The table on the right shows the number of hectares of GMO crops in some of these countries.
- a. Graph the data using a bar graph.
  - b. Summarize your results. What further information would help you compare the data?
  - c. Name a few large countries that are not included in the table. Why do you think they are not included?
- T/I** 6. Researchers have calculated that, on average, each cigarette reduces a person's life by 5.5 min (as a result of the toxic chemicals in cigarettes, many of which are mutagens). If there are 20 cigarettes to a package and an individual smokes half a pack per day, how much will this individual's life have been reduced after one year?
- A** 7. Officials in charge of catching those who use illegal, performance-enhancing drugs in sport are concerned that, in the future, "gene doping" may become a problem. Suggest what this process might be and why athletes might try it.
- K/U** 8. What is a mutation? Would a mutation that caused a deer to be albino be beneficial, harmful, or neutral? If the deer lives in a zoo, would your answer change? Explain.

Use of GMO Crops\*

Country	GMO Crops (hectares)
Argentina	19 101 000
Australia	100 000
Brazil	14 973 000
Canada	7 001 000
China	3 804 000
India	6 192 000
Mexico	100 000
Paraguay	2 590 000
Philippines	300 000
South Africa	1 781 000
Spain	100 000
Uruguay	486 000
United States	57 708 000

\*Among countries with more than 90 000 hectares of GMO crops; rounded to the nearest thousand  
Clive James/International Service for the Acquisition of Agri-Biotech Applications