unit

30 C

Cell Division, Genetics, and Molecular Biology

Cancer is a broad group of diseases associated with the uncontrolled, unregulated growth of cells. Much more active than normal cells, cancer cells divide at rates that far exceed those of the parent cells from which they arose. Cancer cells also do not mature into specific cell types, as do normal cells. Cancer cells cannot carry out some of the functions of normal cells, which in turn can seriously affect a patient's health.

Cancer research aims at understanding how cells become cancer cells, and how they differ from normal cells. A research team at the University of Alberta, led by Dr. Mark Glover, is making significant contributions to our knowledge of one form of breast cancer. People at risk of developing this form of breast cancer have a mutation in a particular gene, which in turn directs the production of a mutant protein. Dr. Glover's group created the first three-dimensional model of the part of this protein that is involved in cancer development. This knowledge may lead to a method to screen patients for this type of cancer early on.

As you progress through the unit, think about these focusing questions:

- What cellular processes allow for reproduction and growth of an organism?
- What regulates the transmission of genetic information from one generation to the next?
- How is DNA responsible for the production of proteins?

UNIT 30 C PERFORMANCE TASK

Investigating Human Traits

Genetics allows us to understand and predict the inheritance of traits. This kind of information can be very important for traits that cause health problems, such as cancer. How can human genetic traits be investigated? What do the patterns of inheritance of some common traits tell us about the genes that determine those traits? At the end of this unit, you may apply your skills and knowledge to complete this Performance Task.

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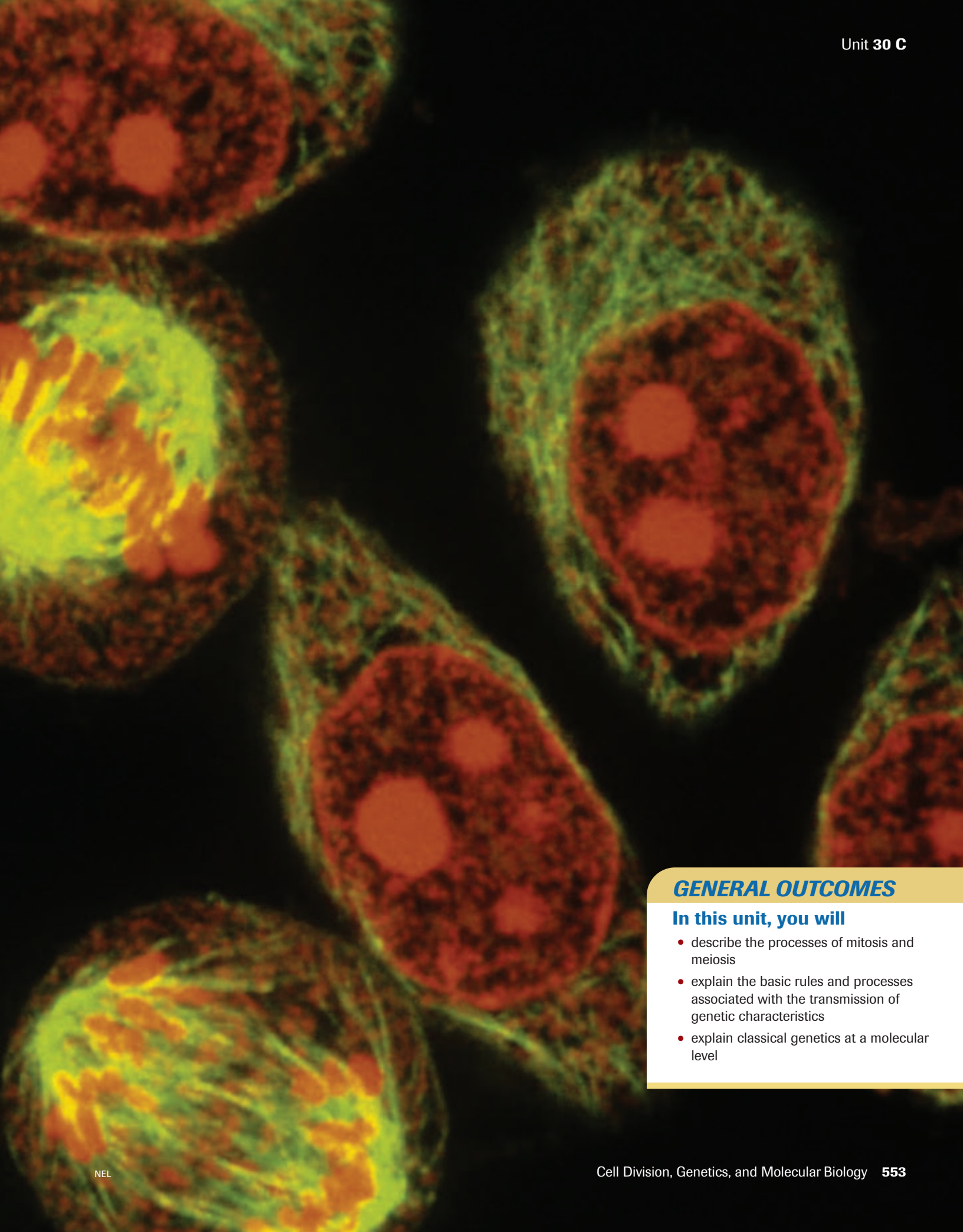

GENERAL OUTCOMES

In this unit, you will

- describe the processes of mitosis and meiosis
- explain the basic rules and processes associated with the transmission of genetic characteristics
- explain classical genetics at a molecular level

Unit 30 C
Cell Division, Genetics, and Molecular Biology

ARE YOU READY?

These questions will help you find out what you already know, and what you need to review, before you continue with this unit.

Prerequisites

Concepts

- DNA, genes, chromosomes
- sexual reproduction
- asexual reproduction
- adaptations and variations
- traits
- nature versus nurture

Skills

- relate biological diversity to genetic diversity
- probability

You can review prerequisite concepts and skills on the Nelson Web site and in the Appendices.

A Unit Pre-Test is also available online.

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Knowledge

1. Identify the cell structures shown in **Figure 1** and explain the importance or function of each.

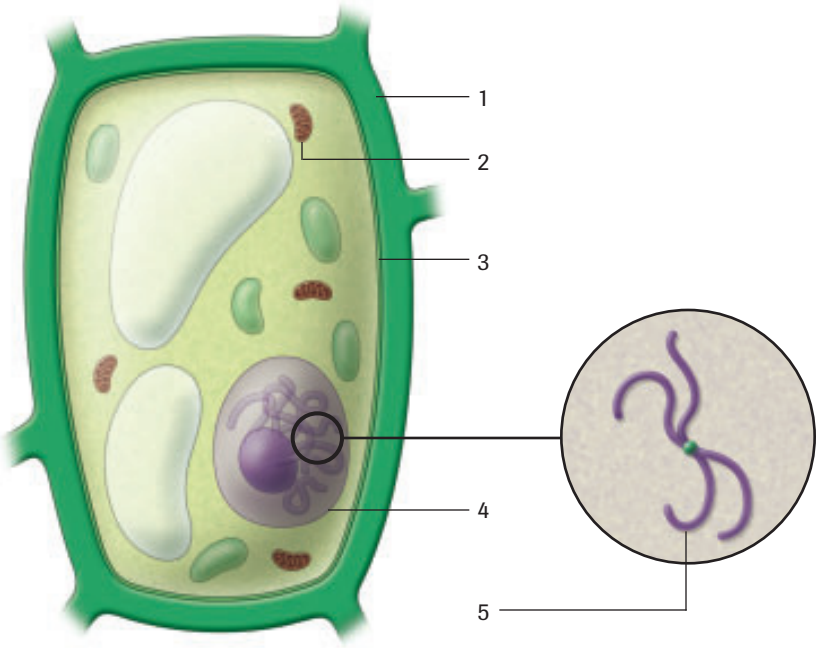


Figure 1

2. (a) Organize the following structures from largest to smallest: organ, chromosome, organism, nucleus, tissue, DNA molecule, cell, gene.
   (b) Copy **Figure 2**. Use the listed structures in (a) as labels for your diagram.

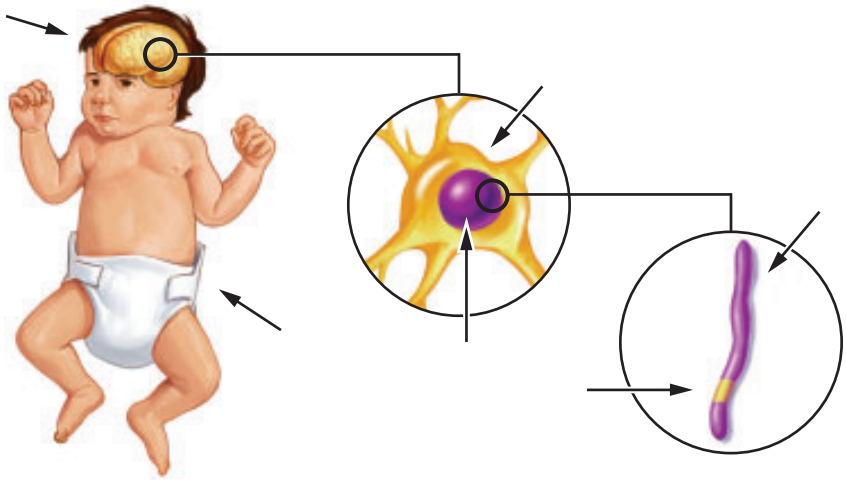


Figure 2

3. If a human muscle cell contains 46 chromosomes, indicate the number of chromosomes that you would expect to find in the cells shown in **Figures 3**, **4**, **5**, and **6**, on the next page.

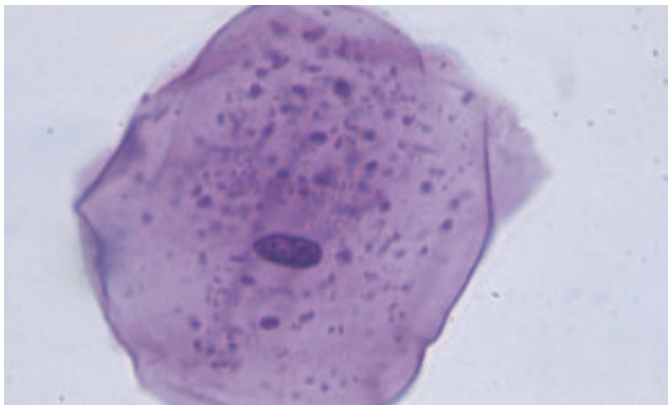


Figure 3
Skin cell, 450×

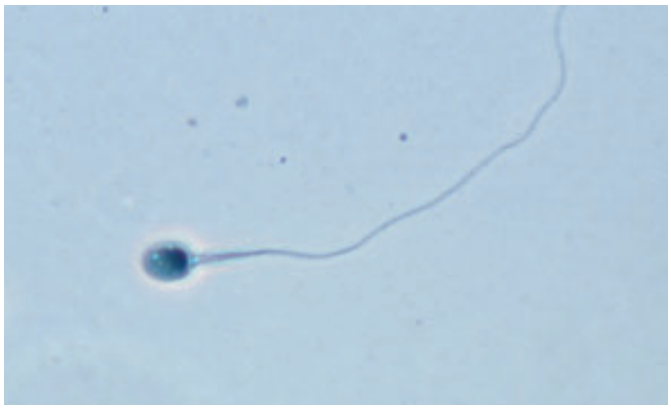


Figure 4
Sperm cell, 1000×

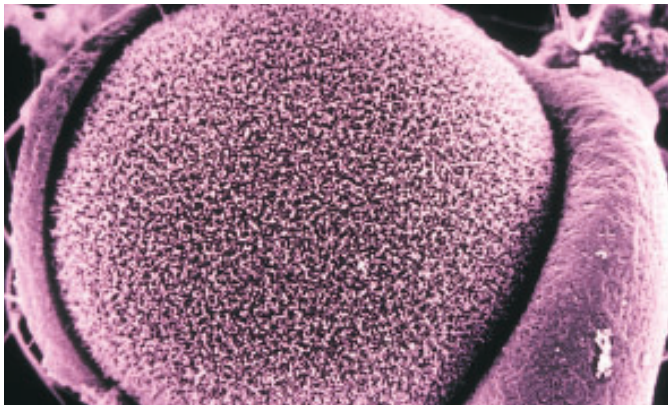


Figure 5
Unfertilized egg cell, 2000×

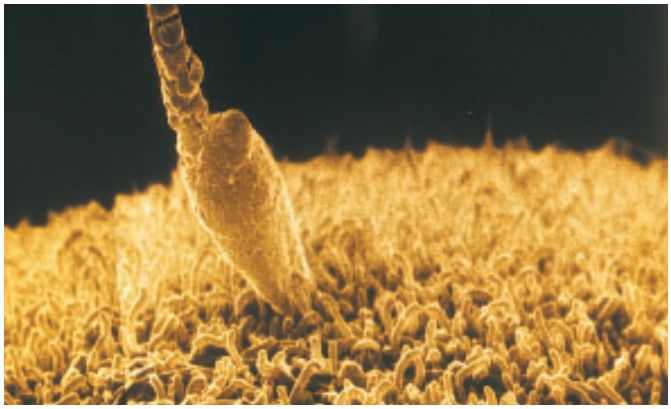


Figure 6
Egg cell being fertilized by sperm cell, 5000×

4. Provide examples of hereditary traits that are
   (a) determined by genes
   (b) influenced by the environment

5. Many single-cell organisms divide by a process called binary fission. One cell divides into two cells identical to each other and identical to the original cell. More complex organisms form specialized sex cells. When sex cells combine from two different organisms, they form a fertilized egg or zygote.
   (a) Identify one advantage of binary fission as a means of reproduction.
   (b) Identify and explain an advantage of reproduction by the union of sex cells from different individuals.

6. Explain why the duplication of genetic material is essential prior to division.

Skills

7. **Table 1** shows the events in a typical cell cycle. Draw and label a circle graph to represent the data.

8. A couple are expecting their third child. After the birth of two boys, they reason that the next child will be a girl.
   (a) Determine the probability of having three boys in a row.
   (b) Determine the probability that the next child will be a girl.

Table 1 Events in the Cell Cycle

Event	Time (h)
rapid growth	15
growth and DNA replication	20
preparation for division	10
mitosis	5

chapter

17 Cell Division

In this chapter

- Exploration: Observing *Daphnia*
- Investigation 17.1: Frequency of Cell Division
- Mini Investigation: Cloning from a Plant Cutting
- Explore an Issue: The Ethics of Stem Cell Research
- Web Activity: Stem Cell Cord Blood
- Investigation 17.2: Identification of a Cancer Cell
- Mini Investigation: Gamete Formation in Grasshoppers
- Investigation 17.3: Comparing Mitosis and Meiosis
- Web Activity: Comparing Life Cycles of Plants
- Web Activity: Dr. Renée Martin
- Web Activity: Modelling Mitosis and Meiosis

All life depends on the ability to grow and reproduce. Both these processes involve cell division. Organisms that reproduce asexually produce offspring that are identical to the parents. Sexually reproducing organisms exchange genetic information, so that the offspring have a unique combination of traits. The genetic material determines the proteins that make up cells, which ultimately give rise to physical traits.

Daphnia (**Figure 1**, next page) is a truly remarkable animal. Females can produce offspring without a mate since they can produce eggs that require no fertilization. Upon development, these eggs become females, which in turn produce females, all of which are identical to each other and to the parent. Then, in response to some environmental cue, *Daphnia* begin producing eggs that develop as either males or females. The males and females produce sex cells. Sexual reproduction occurs when the sperm cells fertilize the egg cells, producing many offspring with a variety of traits. Asexual reproduction occurs when food is plentiful, while sexual reproduction is triggered during times of environmental stress.

All of the cells in *Daphnia* arise from one single cell. To develop into the complex organism in **Figure 1**, that single cell must divide many times. In this chapter, you will explore the events that occur during cell division in order to produce cells of the body and specialized cells involved in reproduction.

STARTING Points

Answer these questions as best you can with your current knowledge. Then, using the concepts and skills you have learned, you will revise your answers at the end of the chapter.

1. Make a list of the advantages of being multicellular.
2. Suggest possible advantages of reproducing
   (a) asexually
   (b) sexually
3. If 22 chromosomes are found in the muscle cell of a mouse, predict the number of chromosomes found in each cell of the following types:
   (a) brain cell
   (b) sperm cell
   (c) fertilized egg cell
   Explain your predictions.

Career Connection: Geneticist

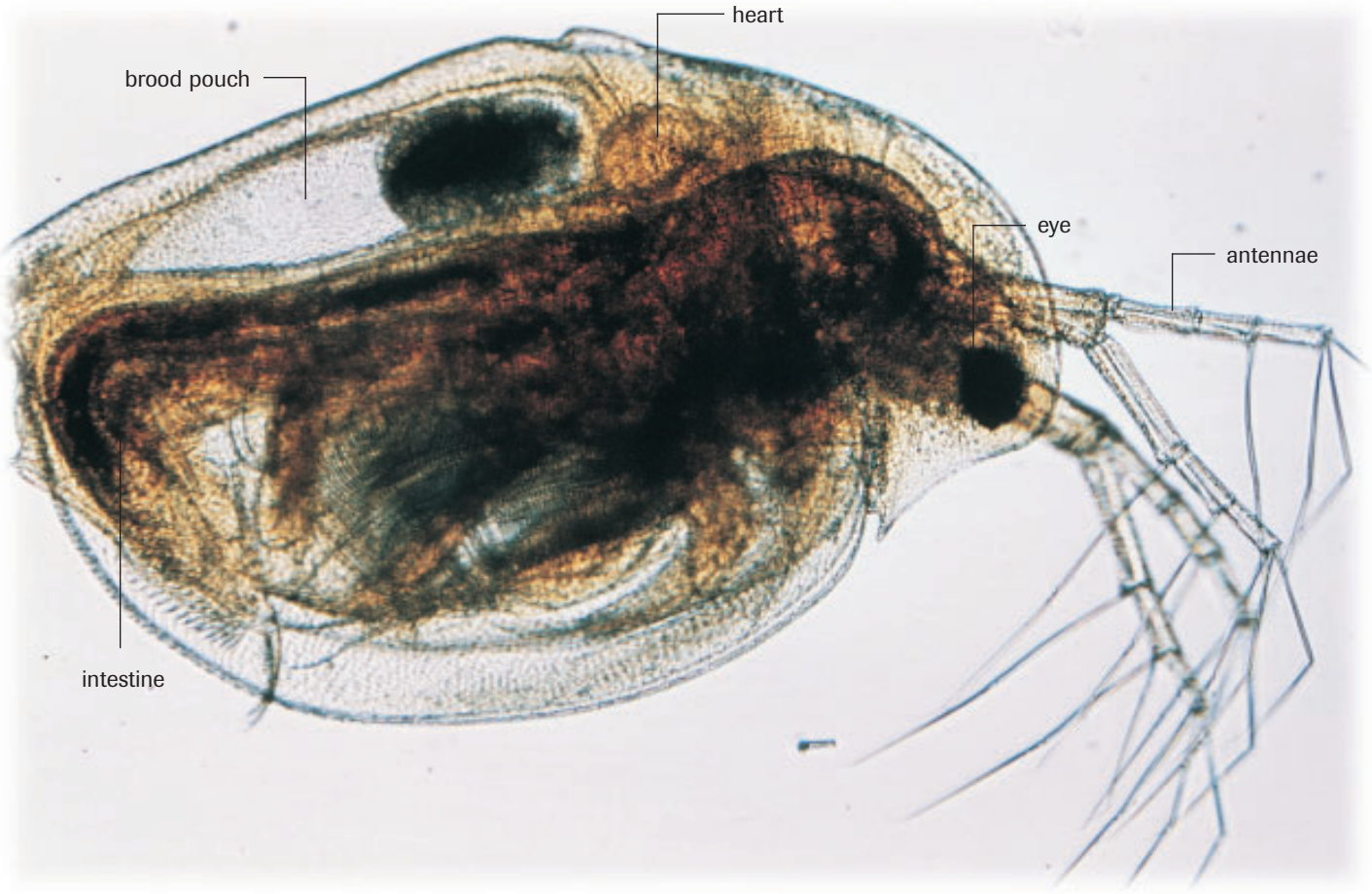


Figure 1
Daphnia is also known as a water flea, but it is a crustacean, not an insect.

Exploration Observing Daphnia

Materials: prepared slide of *Daphnia*, concave depression slide, glycerin, cover slip, *Daphnia* culture, medicine dropper, microscope, ice cubes, cotton swab

- If available, look at a prepared slide of *Daphnia*. Take note of the *Daphnia*'s general appearance and the location of certain features (e.g., eyes, antennae, heart) so that you will be able to identify them more easily in the *Daphnia* culture.
- Remove the prepared slide. Obtain the other materials. Using a cotton swab, smear some glycerin into the depression on the slide. Then, using a medicine dropper, place a small drop of *Daphnia* culture onto the glycerin. Prepare a wet mount by adding a cover slip. Examine the slide under low-power magnification. Pay attention to the movement and heart rate of the organism.
- Place the slide on an ice cube for 3 min, then dry the bottom of the slide with a paper towel and observe once again under low-power magnification.

(a) Why did you smear glycerin on the slide?
(b) Why did you put the slide on an ice cube?
(c) Make and label a scientific drawing of a *Daphnia*.
(d) Do you think that *Daphnia* are composed of many cells? Describe any features that you observe that demonstrate this fact.
(e) Try viewing the *Daphnia* under medium power. (*Hint:* You may have to adjust the diaphragm.) Draw what you see.

17.1 The Cell Cycle

Learning Tip

DNA, the cell's hereditary information, is found in the chromosomes of a cell. In eukaryotic cells (cells with a nucleus), the chromosomes are found in the nucleus. Review this information in Section 6.5 of this book.

All the estimated 100 trillion cells that make up your body arose from a single fertilized egg. As with the frog egg shown in **Figure 1**, this fertilized egg cell underwent a series of divisions that increased the number of cells, thus increasing the size and complexity of your body until eventually you reached your current size. Cell division also maintains a fully grown individual. All multi-cellular eukaryotic organisms grow in size and maintain the cells of their body (the somatic cells) by a sequence of events called the **cell cycle**.

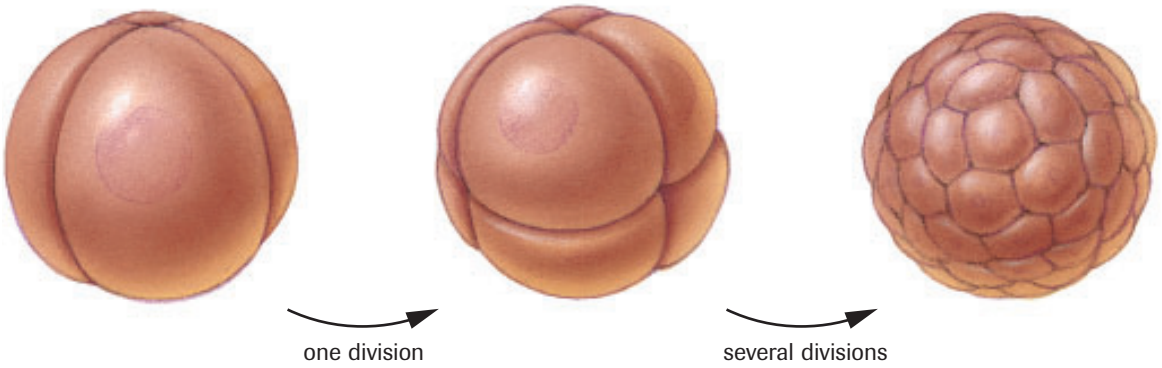


Figure 1
Early stages of cell division of a fertilized frog egg

cell cycle the sequence of stages through which a cell passes from one cell division to the next

mitosis (M) a type of cell division in which a daughter cell receives the same number of chromosomes as the parent cell

cytokinesis the division of cytoplasm

interphase the time interval between nuclear divisions when a cell increases in mass, roughly doubles the cytoplasmic components, and duplicates its chromosomes

The cell cycle is often described as taking place in phases (**Figure 2**, next page). However, the cycle is a continuous process and does not pause after each phase. During the division phase (**mitosis**, or **M**), the components of the cytoplasm and the components of the nucleus of the parent cell are divided to give rise to two identical daughter cells by two processes, mitosis and cytokinesis. Mitosis ensures the equal distribution of the nuclear contents. This process includes the duplication of chromosomes, so that each daughter cell ends up with the same number of chromosomes as the parent cell. **Cytokinesis** divides the cytoplasm and its constituent organelles of the parent cell roughly equally between the daughter cells.

For most cells, the nuclear division that occurs during mitosis marks only a small part of their cycle. The stage between division phases, called **interphase**, is marked by a period of rapid growth (gap 1, or G1), the duplication of chromosomes (synthesis, or S), another period of growth (gap 2, or G2), and preparation for further divisions. Cells carry out their particular functions during interphase.

Chromosome Structure

Before looking at the details of mitosis, you will need to know something about the structure of chromosomes. In animals such as humans, the DNA is divided among a number of chromosomes. Chromosomes contain both DNA and a number of proteins.

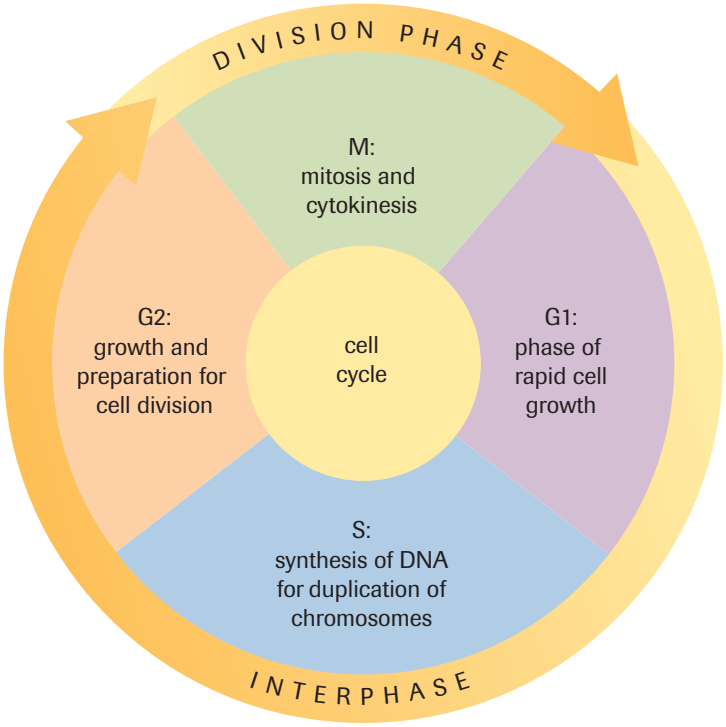


Figure 2
The cell cycle. The circle represents the entire life cycle of the cell, which can be divided into two major phases: interphase and the division phase. Most cells spend the majority of their time in interphase.

This combination of DNA and proteins is called **chromatin**. As the cell moves through the cell cycle, chromosomes may be either uncondensed or condensed. Uncondensed chromosomes are long, thin strands that cannot be seen under a light microscope. A condensed chromosome can be seen under a light microscope and may resemble the diagram in **Figure 3**. Condensed chromosomes may be either unduplicated or duplicated. In a duplicated chromosome, the original chromosome and its duplicate are attached to each other by a structure called the **centromere**. While attached to one another, the two chromosome duplicates are called **sister chromatids**. Since sister chromatids contain identical genetic information, the pair, attached at the centromere, is still considered to be one chromosome.

chromatin the complex of DNA and protein that make up chromosomes

centromere the structure that holds chromatids together

sister chromatids a chromosome and its duplicate, attached to one another by a centromere until separated during mitosis

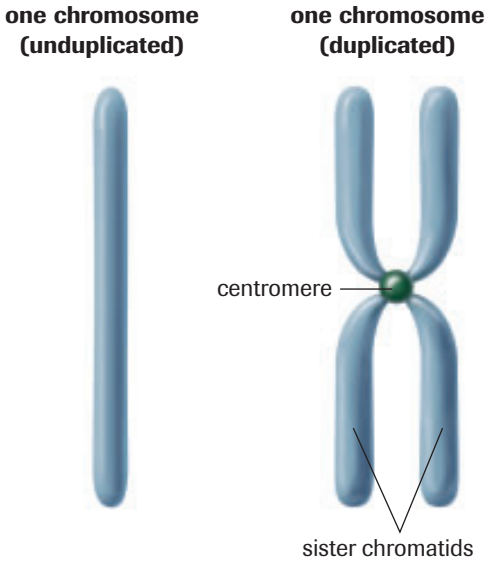


Figure 3
An unduplicated and a duplicated chromosome

Interphase

Cells spend most of their lives in interphase. In this phase of the cell cycle, cells are not actively dividing. Interphase includes the G1, S, and G2 phases of the cell cycle. Cells in interphase grow and undergo the various metabolic processes needed for their functioning during G1, S, and G2.

Chromosomes are uncondensed throughout interphase (**Figure 4**). During G1, cells undergo a period of rapid growth, and the chromosomes are unduplicated. During the S phase, cells begin to prepare for division during interphase by duplicating its chromosomes. At the end of the S phase, all the chromosomes are therefore duplicated chromosomes. During G2, the cell again grows and it completes the preparations for division (mitosis, or the M phase).

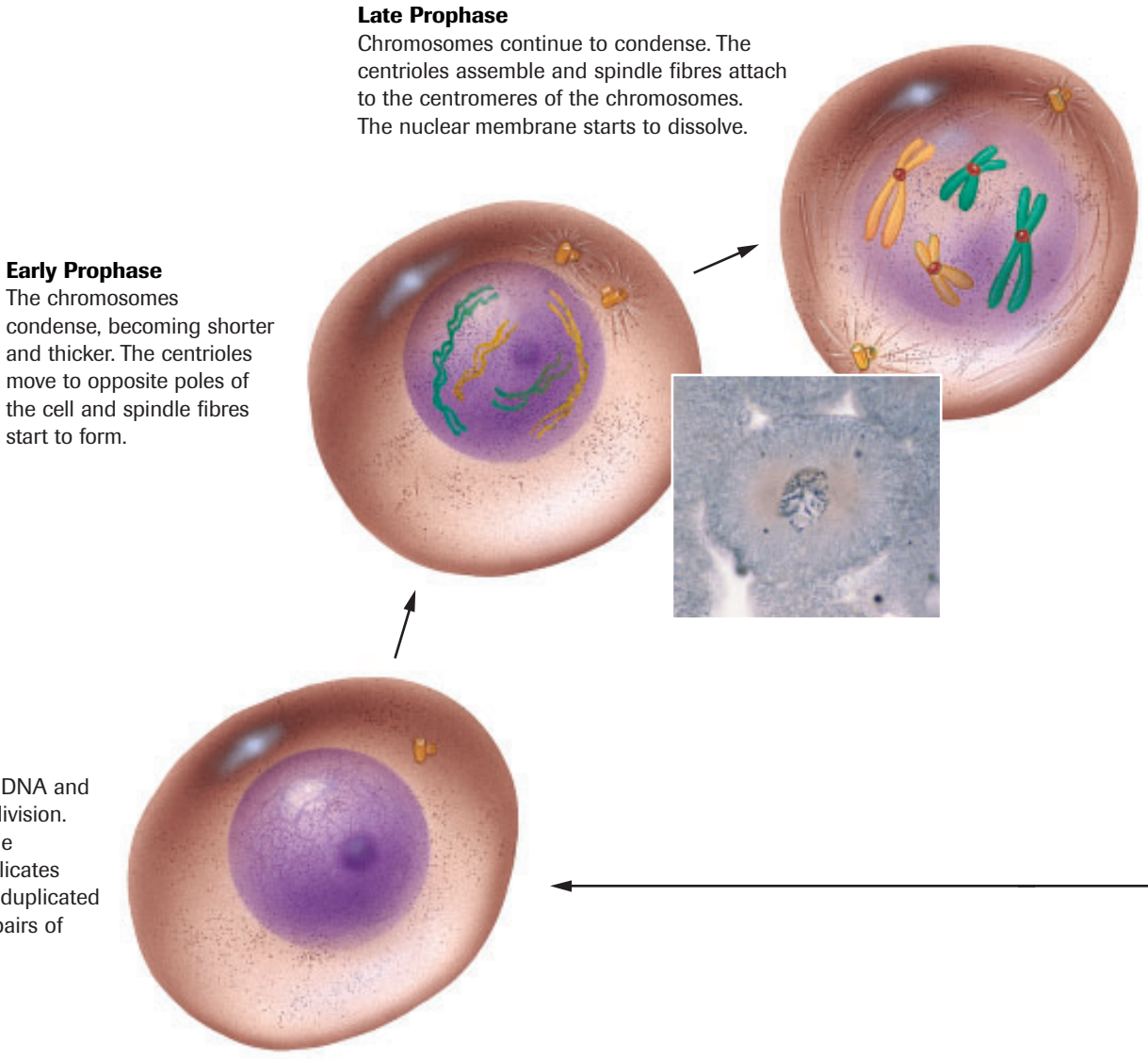


Figure 4
Interphase and mitosis in an animal cell. Interphase includes the G1, S, and G2 phases of the cell cycle. Mitosis and cytokinesis occur during the M phase.

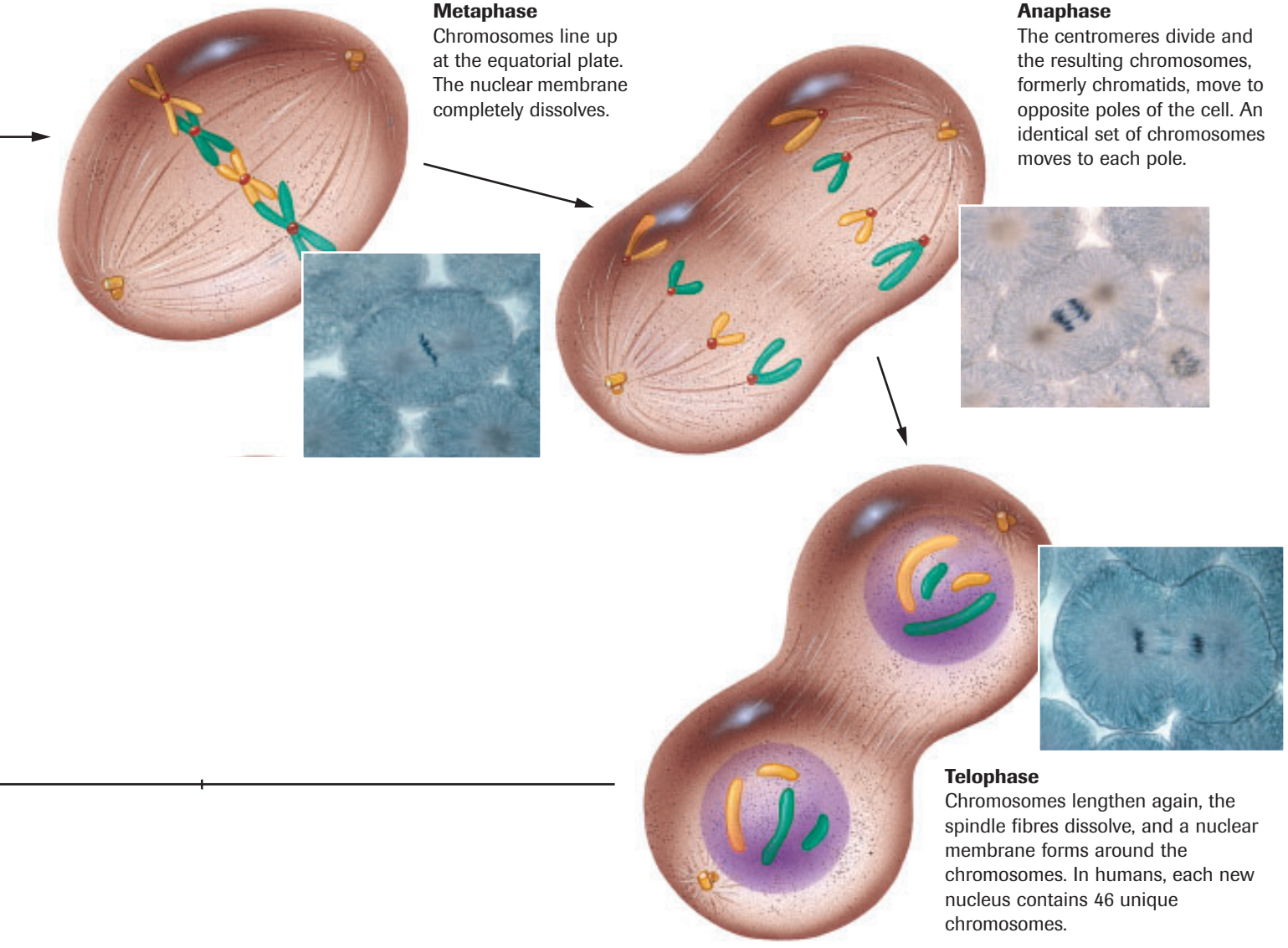
The Stages of Mitosis

Prophase

Prophase is the first phase of mitosis. The chromosomes in the nucleus become visible under a microscope as they shorten and thicken (**Figure 4**). In animal cells, a small body in the cytoplasm separates and its parts move to opposite poles of the cell as the chromosomes become visible. These tiny structures, called **centrioles**, provide attachment for the **spindle fibres**, which serve as guide wires for the attachment and movement of the chromosomes during cell division. Collectively, the centrioles and spindle fibres make up the spindle apparatus. Most plant cells do not have centrioles, but spindle fibres still form and serve a similar purpose. The centromere joining the two chromatids helps anchor the chromosomes to the spindle fibres. When viewed under a microscope during prophase, the nuclear membrane appears to fade; in effect, it is dissolving to allow the separation of chromosomes and cell organelles.

centriole small protein body found in the cytoplasm of animal cells that provides attachment for spindle fibres during cell division

spindle fibre protein structure that guides chromosomes during cell division



Metaphase
Chromosomes line up at the equatorial plate. The nuclear membrane completely dissolves.

Anaphase
The centromeres divide and the resulting chromosomes, formerly chromatids, move to opposite poles of the cell. An identical set of chromosomes moves to each pole.

Telophase
Chromosomes lengthen again, the spindle fibres dissolve, and a nuclear membrane forms around the chromosomes. In humans, each new nucleus contains 46 unique chromosomes.

Metaphase

The second phase of mitosis is metaphase. Chromosomes composed of sister chromatids move toward the centre of the cell. This centre area is called the equatorial plate, because, like the equator of Earth, it is midway between the poles of the cell. The chromosomes appear as dark, thick filamentous structures that are attached to the spindle fibres. Even though they are most visible at this stage, it is still very difficult to count the number of chromosomes in most cells because the chromosomes are entangled. Chromatids can become intertwined during metaphase.

Anaphase

Anaphase is the third phase of mitosis. The centromeres divide and the sister chromatids, now referred to as chromosomes, move to opposite poles of the cell. If mitosis proceeds correctly, the same number and type of chromosomes will be found at each pole. Occasionally, segments of the chromatids will break apart, and may reattach, in anaphase.

Telophase

The last phase of mitosis is telophase. The chromosomes reach the opposite poles of the cell and begin to lengthen. The spindle fibres dissolve and a nuclear membrane forms around each mass of chromatin. Telophase is followed by cytokinesis, the division of the cytoplasm.

Cytokinesis

Once the chromosomes have moved to opposite poles, the cytoplasm begins to divide. Cytokinesis appears to be quite distinct from nuclear division. In an animal cell, a furrow develops, pinching off the cell into two parts. This is the end of cell division. In plant cells, the separation is accomplished by a cell plate that forms between the two chromatin masses. The cell plate will develop into a new cell wall, eventually sealing off the contents of the new cells from each other.

Practice

1. List the stages of mitosis. Briefly describe what occurs in each stage. To help in your description, sketch the sequence of events that occurs in an animal cell. Include labels for different structures.
2. A cell with 10 chromosomes undergoes mitosis. Indicate how many chromosomes would be expected in each of the daughter cells.

A Cell Clock

How old can cells become? If cells continue to undergo mitosis, could an organism stay eternally young and live forever? Research on cultured cells (cells grown in a nutrient medium) indicates that a biological clock may regulate the number of cell divisions available to cells. When immature heart cells maintained in tissue culture were frozen, they revealed an internal memory of the number of cell divisions they had undergone. If a cell had undergone twenty divisions before freezing, the cell completed another thirty divisions once it thawed, then died. When a cell was frozen after ten divisions, it completed another forty divisions after thawing and then died. Cells always completed a total of fifty divisions no matter how long the freezing or at what stage the cell division was suspended (**Figure 5**).

EXTENSION

Mitosis and Cell Division in Plants and Animals

This Audio Clip highlights the observable differences between plant and animal cell mitosis and cytokinesis.

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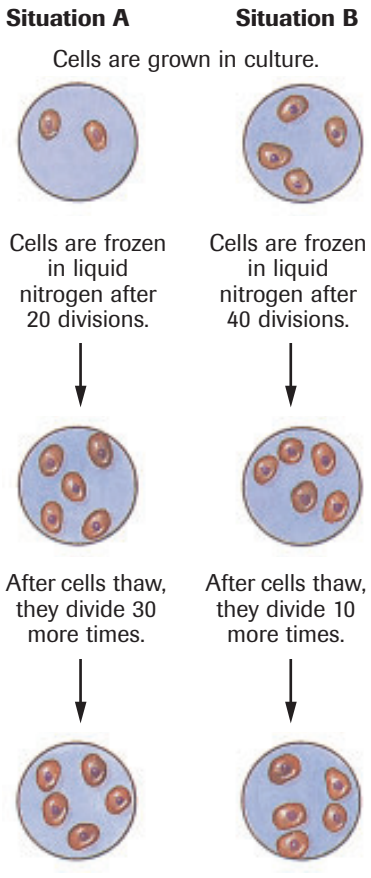

Figure 5
Cell division appears to be controlled by a biological clock.

Not all cells of the body have the same ability to undergo mitosis. Age is one reason cells stop dividing. However, division is usually stopped by cell specialization. Relatively unspecialized cells, such as skin cells and the cells that line the digestive tract, reproduce more often than do the more specialized muscle cells, nerve cells, and secretory cells. Only two cell types in the human body divide endlessly: the sperm-producing cells, called spermatogonia, and the cells of a cancerous tumour. Males are capable of producing as many as one billion sperm cells a day from the onset of puberty well into old age. However, once the sperm cells are formed, they lose the ability to divide further. Cancer cells divide at such an accelerated rate that the genes cannot regulate the proliferation and cannot direct the cells toward specialization.

It would appear that the more specialized a cell is, the less able it is to undergo mitosis. The fertilized egg cell is not a specialized cell; differentiation begins to occur only after its third division, which results in eight cells. Interestingly, it is at the point where differentiation begins that the biological clock within the cell is turned on.

EXTENSION

Cancer and Metastasis

Cells that divide uncontrollably can become cancer. This animation shows how cancer cells can spread from one part of the body to another.

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INVESTIGATION 17.1 ***Introduction***

Frequency of Cell Division

In this activity, you will view and compare cells from onion cells and from a whitefish blastula in various stages of mitosis. Because slides are used, the cell divisions you will be viewing are frozen in time. Therefore, it will not be possible for you to watch a single cell progress through the stages of mitosis. Based on your observations, you will determine the frequency of cell division and construct a clock representing the division cycle, given the time taken to complete one cycle of mitosis. In a table, you will record the number of cells in each stage of mitosis.

Report Checklist

● Purpose	○ Design	● Analysis
○ Problem	○ Materials	● Evaluation
○ Hypothesis	○ Procedure	● Synthesis
○ Prediction	● Evidence	

To perform this investigation, turn to page 587.

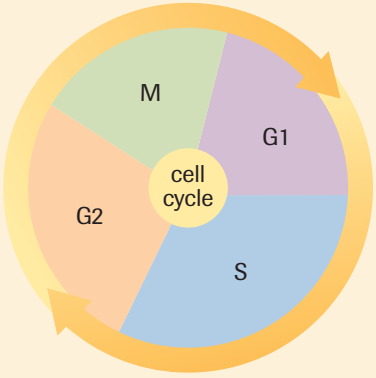
SUMMARY *The Cell Cycle*

- Cell division produces new cells for cell growth and for the replacement of worn-out cells in the body.
- Cell division involves a series of steps that produce two genetically identical daughter cells. Two divisions occur during cell division: nuclear division (mitosis) and cytoplasmic division (cytokinesis).
- During interphase, genetic material is replicated.
- Cells seem able to divide only a finite number of times.
- Cells lose the ability to divide as they specialize.

Section 17.1 Questions

1. During interphase, what event must occur for the cell to be capable of undergoing future divisions?
2. Using a dictionary, look up the meaning of the prefixes used in the stages of mitosis: *pro-, meta-, ana-,* and *telo-*. Why would they be used in the naming of the phases of mitosis?
3. Compare and contrast the structure of the daughter cells with that of the original parent cell.
4. Describe the structure and explain the function of the spindle fibres.
5. What is the significance of cytokinesis? Speculate what would happen if cytokinesis did not occur.
6. When a cell has reached its maximum size, what two alternatives does it have? When does the cell carry out one alternative over the other?
7. What would happen if you ingested a drug that prevented mitosis? What if it only prevented spindle fibre formation?
8. A cell from a tissue culture has 38 chromosomes. After mitosis and cytokinesis, one daughter cell has 39 chromosomes and the other has 37. What might have occurred to cause the abnormal chromosome numbers?
9. Suppose that during mitosis, both sister chromatids moved to the same pole, resulting in daughter cells with a different number of chromosomes than the parent cell. How might this abnormality affect cell structure, cell function, or both?
10. Explain the concept of the cell clock.
11. Suggest reasons why skin cells, blood cells, and the cells that line the digestive tract reproduce more often than other types of cells such as muscle cells. If some of these cells were to become cancerous, how might a chemical therapy to stop those cells from reproducing work?
12. (a) Describe the differences between the two cell cycles in **Figure 6**.
    (b) Which cell cycle do you believe would represent a cell of an embryo and which would represent an unspecialized cell in an adult? Give your reasons.
13. List areas of the body where you think cell division is most rapid. Also, indicate the comparative level of specialization of the cells in each area. Explain your predictions.
14. It is believed that weed killers like 2,4-D and 2,4,5-T may work by stimulating cell division. Why would the stimulation of cell division make these chemicals effective weed killers?
15. At one time, blood was transfused only from younger individuals to the elderly. It was believed that younger blood would provide the elderly with more energy. Do older people actually have older blood cells? Support your answer.
16. X-rays and other forms of radiation break chromosomes apart. Physicians and dentists will not X-ray pregnant women. Even women who are not pregnant wear a lead apron when being X-rayed near the reproductive organs. The apron blocks the passage of X-rays. Why is it undesirable to X-ray the reproductive organs? Why is it especially undesirable to X-ray pregnant women?
17. Scientists have developed techniques aimed at getting highly-specialized cells to act as if they are immature cells that have not yet become specialized. Why would scientists want to be able to get a mature nerve cell to respond like a cell that hasn't undergone specialization?

Cell Cycle for Cell A: 36 h



Cell Cycle for Cell B: 25 h

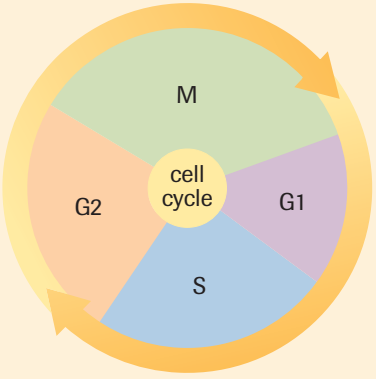


Figure 6

Applications of the Cell Cycle 17.2

Scientists continue to study the cell cycle and to gain a deeper understanding of the mechanisms and the role of the process. As more is learned about the cell cycle, we have been able to apply this knowledge to many human needs. There are various perspectives on the costs and benefits of these new technologies, and when they are appropriate to use.

Cloning

Cloning is the process of forming identical offspring from a single cell or tissue in the parent organ. A clone originates from a single parent cell, and both the clone and parent have identical (or nearly identical) nuclear DNA. Although some clones show accidental changes in genetic information, cloning does not result in the variation of traits that would occur with the combination of male and female sex cells. Cloning is therefore considered a form of asexual reproduction. In fact, clones occur naturally. Some species, such as hydra (**Figure 1 (a)**) reproduce by undergoing mitosis to produce buds with identical DNA to the larger parent cell. The smaller plantlets on a runner of a strawberry plant are identical clones of the larger parent plant (**Figure 1 (b)**). In animals, offspring with an identical genetic makeup are sometimes produced when a single fertilized egg undergoes mitosis and the resulting early embryo (called a zygote) then splits in two (**Figure 1 (c)**). This results in identical twins. They are also called monozygotic twins, since they formed from a single zygote. Fraternal twins are formed when two different eggs are fertilized separately. They are also known as dizygotic twins. Fraternal twins, therefore, are no more genetically similar than are non-twin siblings (**Figure 1 (d)**).

DID YOU KNOW ?

Multiple Births

It has been estimated that 1 in 85 births will produce twins, 1 in 7500 will produce triplets, 1 in 650 000 will produce quadruplets, and 1 in 57 000 000 will produce quintuplets.

(a) **(b)** **(c)** **(d)**

Figure 1

(a) Hydra reproduce asexually by budding. The buds break off to form separate, genetically identical organisms.

(b) The strawberry plant can reproduce asexually by forming genetically identical plantlets on runners.

(c) Identical twins originate from a single fertilized egg that undergoes mitosis to produce an early embryo which then splits into two, producing two genetically identical individuals.

(d) Development of fraternal twins does not involve the splitting of a fertilized egg. Instead, fraternal twins develop from two independent fertilization events, such as occurs when a mother has two eggs in her uterus that are fertilized by two different sperm cells. Each fertilized egg then develops independently.

mini Investigation — Cloning from a Plant Cutting

In some plants, asexual reproduction is accomplished naturally when a portion of the plant, such as a stem or leaf, breaks off and develops roots at the base of the broken portion. It is possible for the broken part to become a new plant. This activity is an example of artificial propagation.

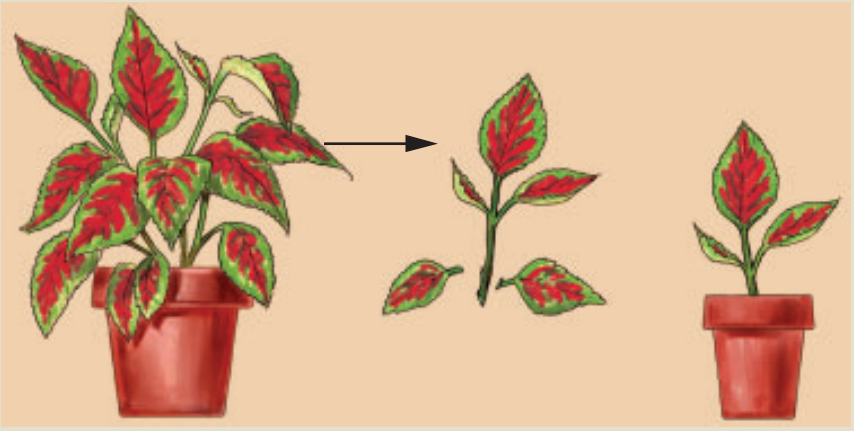
Materials: coleus plant, scissors, goggles, gloves, fungicide, flower pot, potting soil, apron

The fungicide is poisonous. Review the MSDS before beginning this investigation. Any spills on the skin, in the eyes, or on clothing should be washed immediately with cold water. Report any spills to your teacher.

- Perform the following steps as shown in **Figure 2**.
  1. Using scissors, carefully cut off the tips of three coleus stems. Cut on an angle. Include several leaves on each stem.
  2. Remove a few leaves from the bottom. Put on splash goggles, and wear gloves and/or use tongs to immerse the stem in fungicide.
  3. Plant the cuttings in soil.
- Record each cutting's initial height and number of leaves. Take these measurements every week for two months.
- Describe the new plants each time.

(a) What evidence suggests that coleus can regenerate parts of the plant that were lost?

(b) Without removing the plant from the pot, how can you demonstrate that the roots from the cutting are growing?

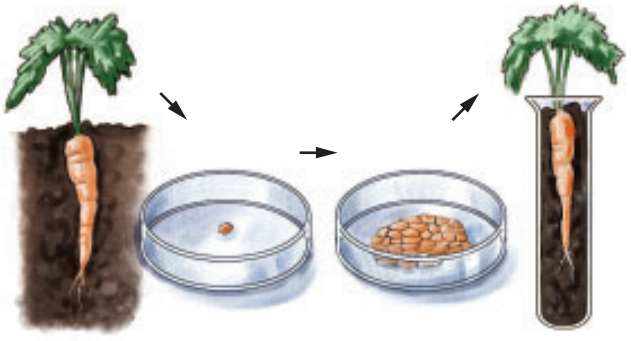


step 1 step 2 step 3

Figure 2

Plant Cloning Technology

In 1958, Fredrick Stewart created great excitement in the scientific world when he revealed that he had produced a plant from a single carrot cell (**Figure 3**). Today, this technique



single cell extracted from carrot carrot clone

Figure 3
Fredrick Stewart was able to grow a clone from a single cell of a carrot plant. This allowed production of many identical individuals from a sexually reproducing species. This was the first application of knowledge of mitosis in generating clones.

is commonly called cloning. Many commercially important plant species, including orchids, are now produced from clones. Unlike plants that arise from sexual reproduction, cloned plants are identical to their parents. This allows production of strains of plants with predictable characteristics. Not all plant species can be cloned, however. Carrots, ferns, tobacco, petunias, and lettuce respond well to cloning, but the grass and legume families do not. Scientists continue to investigate these differences.

Each cell in the cloned plant contains the complete complement of chromosomes from the parent. As the new plant develops, it undergoes mitosis to increase in size. Some cells then specialize (differentiate) and form roots, stems, or leaves, until a complete plant is formed.

Animal Cloning Technology

While plant cloning experiments were being conducted, Robert Briggs and Thomas King were busy investigating nuclear transplants in frogs. Working with the common grass frog, the scientists extracted the nucleus from an unfertilized egg cell by inserting a fine glass tube, or micropipette, into the cytoplasm and sucking out the nucleus (**Figure 4**). A cell without a nucleus is referred to as **enucleated**.

enucleated the condition where a cell does not contain a nucleus

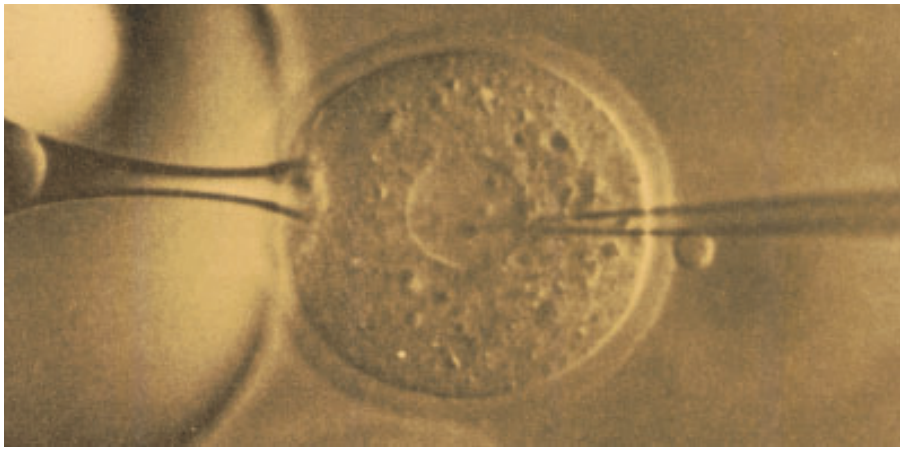


Figure 4
A small glass tube, called a micropipette, is used to remove the nucleus from a cell and later introduce a new nucleus.

Next, the nucleus of a cell from a frog embryo in the blastula stage of development was removed and inserted into the enucleated cell (**Figure 5**). The egg cell with the transplanted nucleus began to divide much like any normal fertilized egg cell. In later trials, the cell with the transplanted nucleus occasionally grew into an adult frog. The adult frogs displayed the characteristics from the transplanted nucleus. Careful analysis proved that the adults were clones of the frog that donated the nucleus.

However, different results were obtained when the nucleus was taken from cells at later stages of development. For example, the nucleus from cells in a later stage, called the gastrula stage, did not bring the enucleated egg from the single-cell stage to the adult. If mitosis occurred at all, it did not progress as far as it did in eggs that received a blastula nucleus. The difference is that the nucleus of a cell in the gastrula stage of development, unlike a cell in the earlier blastula stage, has specialized. As cells begin to specialize, they become less able to undergo mitosis.

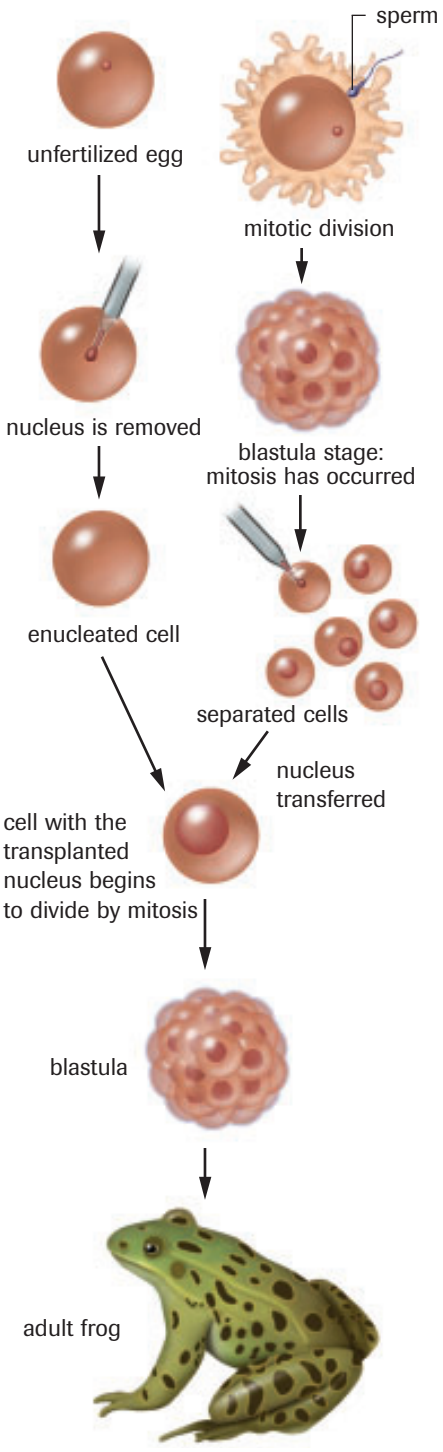


Figure 5
Cloning a common grass frog using embryo splitting

Cloning from adult mammalian cells has proved even more difficult, since they tend to be highly specialized. Until recently, the only way to get clones was by splitting off cells from a developing embryo (**Figure 6**). However, cells beyond the eight-cell stage of development seem to be unable to stimulate cell division.

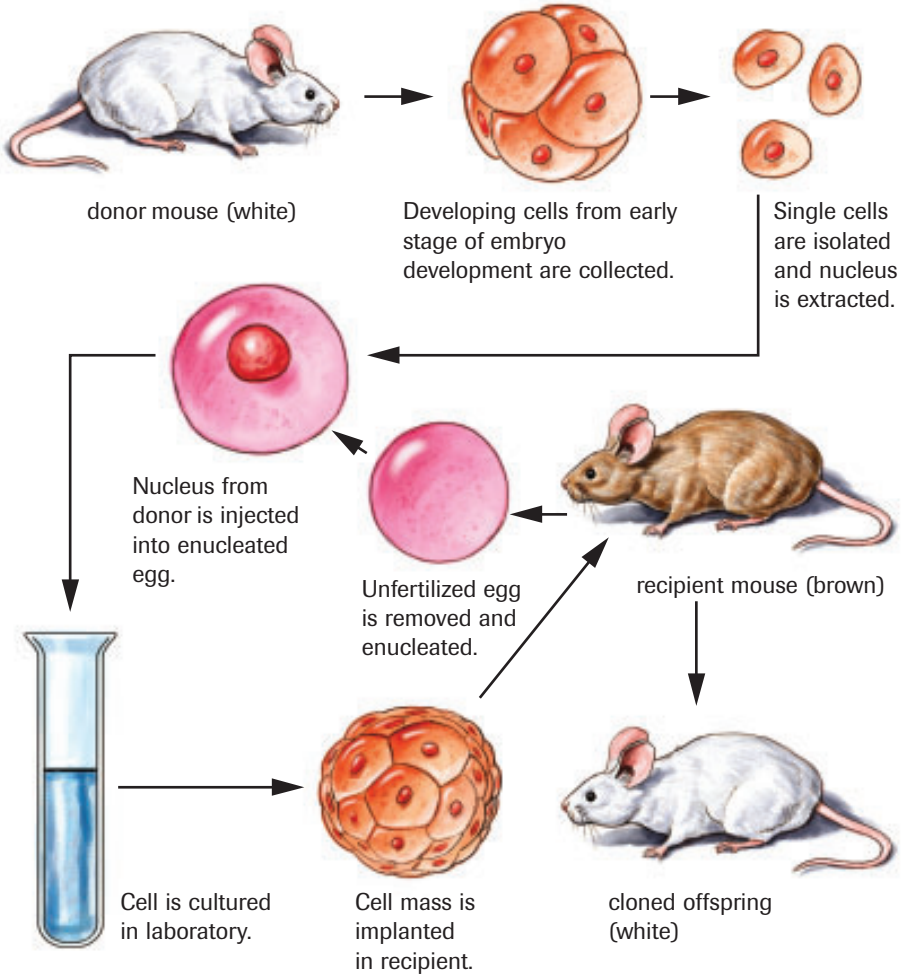


Figure 6
Cloning a mammal using embryo splitting

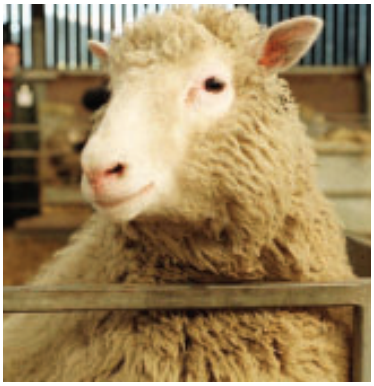


Figure 7
Dolly could claim three different sheep as mothers. The genetic mother died before Dolly was born.

The long-held scientific belief that adult cells cannot be used to clone animals was disproved with the appearance of a sheep named Dolly. Dr. Ian Wilmut, of the Rosalind Institute in Scotland, extracted the nucleus from an udder cell of an adult Finn Dorsett sheep and placed the nucleus into the enucleated egg cell from a Poll Dorsett sheep. The egg was allowed to develop in a Petri dish until an early embryo stage was reached. Then this embryo was placed into the womb of a third sheep, a Scottish Blackface. Her genetic information was shown to be identical to that of the Finn Dorsett adult; Dolly was a clone (**Figure 7**).

Medical experimentation and research could potentially benefit from the availability of cloned animals. For example, experiments on the effectiveness of a drug are often difficult to interpret because of the genetic variation among the individuals tested. If all the test subjects were genetically identical, clearer results could be obtained. In agriculture, the strongest livestock could be cloned, decreasing farmers' losses due to disease, and thereby increasing yield. However, many people have moral and ethical problems with this technology and worry about the impact on society .

Practice

1. List the steps involved in cloning animals from nuclei taken from the blastula stage of development.
2. Why are identical twins often called "nature's clones"?
3. Do all the cells of your body divide at the same rate? Explain.
4. What is an enucleated cell?

EXTENSION

Stem Cells
This *NOVA* video asks what are stem cells and how do we find a balance between hope for cures and respect for life.

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EXPLORE an issue

Issue Checklist

- Issue
- Design
- Analysis
- Resolution
- Evidence
- Evaluation

The Ethics of Stem Cell Research

A **stem cell** is a cell from which any other type of cell can arise (stem). Upon receiving the appropriate signals, stem cells differentiate into specialized cells with a particular function, such as heart muscle cells. Since a stem cell has not differentiated, it can undergo many cell divisions. Fertilized eggs and early embryos are composed entirely of stem cells. Plants retain many stem cells throughout life, in the growing tips of roots and shoots. Some adult animals also retain many stem cells, such as in salamanders that can grow a lost tail. In contrast, the adult human body has very few stem cells. Stem cells are found in the adult human body in bone marrow, fat, blood, and even in hair follicles. The richest source of non-embryonic stem cells is umbilical cord blood.

Stem cells have the potential of having enormous medical benefits. Since stem cells can potentially give rise to any other type of cell, they may be able to help people whose cells are not able to function properly. For example, stem cells could be used to replace faulty insulin-producing cells in the pancreas of diabetics or faulty neurotransmitter-producing cells in the brains of people with Parkinson disease.

Some people do not agree with the use of stem cells on ethical grounds. Scientists still do not fully understand how a single, unspecialized cell becomes a complex organism with many specialized cells. Some people worry that scientists may use human embryos to answer these questions. Others believe that any cell that can potentially give rise to a human being should not be used for research or therapy.

- In small groups, conduct background research on this rapidly changing field of research using newspapers, periodicals, CD-ROMs, and the Internet. Outline how the issue is changing and any new issues that are emerging. Prepare a bibliography and make notes as you work.

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- Based on your background research, describe one ethical issue related to the use of stem cells in research or therapy.
- For the issue you have stated, write a statement that describes one viewpoint. For example, you might state, "Withholding a potential cure because it uses stem cells is unethical, because it causes people with a medical condition to suffer."
- Decide whether you agree or disagree with the statement. If necessary, conduct additional research to find evidence to support or refute your viewpoint.
- Write a position paper. Be prepared to defend your group's position to your classmates.

WEB Activity

Web Quest—Stem Cell Cord Blood

Research into stem cell cord blood has provided major steps forward in scientific understanding. It is becoming commonplace for parents to save the blood from their newborn's umbilical cord and to bank it in case of future medical needs. The issue is no longer whether or not banking the cord blood is acceptable, but rather the argument between the use of private or public stem cell cord blood banks. This Web Quest asks you to develop a supported position on this issue and create a presentation that can be given to your class.

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EXTENSION

Stem Cells Update
This *NOVA* video discusses a new technique for creating stem cells that may ease ethical concerns.

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telomere the cap at the end of a chromosome

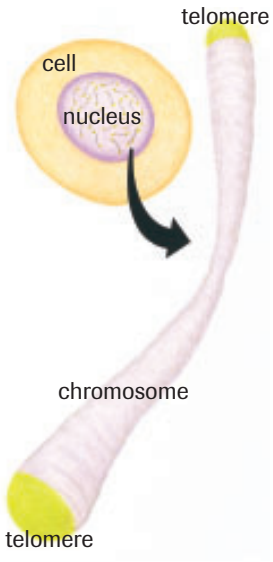

Figure 8
Telomeres are end caps of chromosomes. An enzyme, called telomerase, acts on the telomere causing changes in length.

Mitosis and Telomeres

Telomeres are caps at the ends of chromosomes (**Figure 8**). Scientists have determined that telomeres reduce in length each time a cell goes through the cell cycle and divides. Telomeres might have a role in cell aging and in the behaviour of cancer cells.

In 1984, Carol Greider and Elizabeth Blackburn set out to find the enzyme that affected the length of the telomere. Not only did they find the enzyme, but they also discovered much about how it works. Dr. Blackburn demonstrated a connection between telomerase and aging. Yeast cells that lack the enzyme telomerase undergo telomere shortening and eventually die. Other researchers working in Scotland found that as human cells age, telomere length shortens. The length of the chromosomes of a 70-year-old human is much shorter than that of a child. As we saw in Section 17.1, normal cells pass through the cell cycle only a finite number times. Once a cell can no longer undergo mitosis, cell death occurs. Telomeres length serves as a molecular "clock" for cellular aging.

What impact does telomere length have on cloning technology? The answer is not yet clear. Since Dolly was cloned from the cells of a six-year-old sheep, she began life with shorter telomeres than would a non-cloned sheep. Dolly developed arthritis at an early age and died of lung disease in February of 2003 at only six years of age—half the normal life expectancy of a sheep. These events may be linked to telomere length. However, some cloned animals appear to have longer telomeres, as if they were younger.

In the human body, cells generally undergo mitosis only 50 to 100 times during their lifespan. Cancer cells, however, never seem to lose their ability to divide, and their telomere length is also maintained. Telomerase is also not present in most normal cells. A group working at McMaster University under the direction of Calvin Harley was the first to show that telomerase is reactivated in human cancer cells. This allows cancer cells to maintain telomere length and, therefore, their ability to divide (**Figure 9**). Dr. Harley is now working with a pharmaceutical company to develop a drug that can block telomerase action. They hope that decreasing telomerase activity will slow cell division of the cancer cells, but have little impact on normal cells.

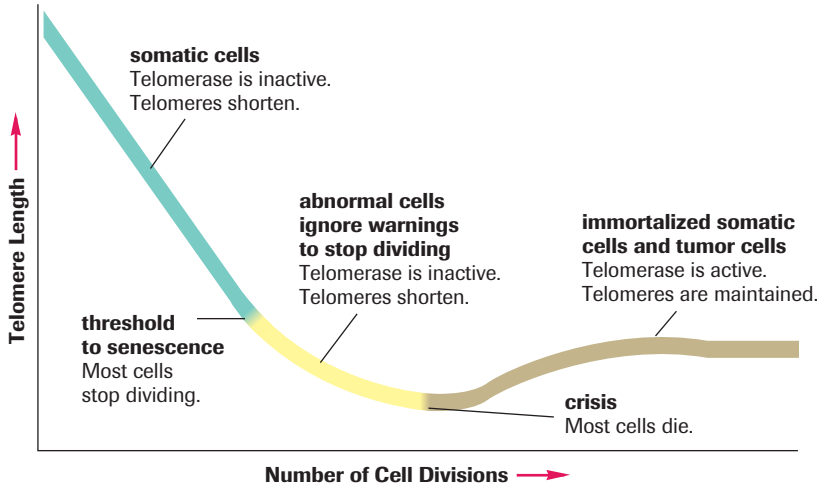

Figure 9
The activity of telomerase in normal cells (turquoise line) decreases as the cell ages. Eventually, the cells reach the point where damage to the chromosomes will result if the telomeres become any shorter. At this point, normal cells stop dividing and die. Abnormal cells continue to divide (yellow line). Cancer cells (brown line) reactivate telomerase and so are able to continue mitosis.

INVESTIGATION 17.2 *Introduction*

Identification of a Cancer Cell

Cancer cells have unique features that can be used to distinguish them from non-cancerous cells. These differences can be used by medical professionals as a means of detecting cancer, often in earlier, easy-to-treat stages by technologies such as X-rays, infrared photography, and cell biopsies. Some of these differences can be viewed using a light microscope. What are these differences? Do they relate to the ability of these cells to continue undergoing mitosis?

Report Checklist

○ Purpose	● Design	● Analysis
● Problem	○ Materials	● Evaluation
○ Hypothesis	○ Procedure	● Synthesis
○ Prediction	● Evidence	

In this investigation, you will examine stained slides of cancerous and non-cancerous cells under a light microscope to observe some differences between these cell types.

To perform this investigation, turn to page 589.

SUMMARY *Applications of the Cell Cycle*

- Cloning is the process of forming identical offspring from a single cell or tissue.
- Cloning permits the production of offspring with characteristics identical to those of the parent.
- Some plants and animals naturally clone themselves (reproduce asexually).
- Technologies have been developed to clone both plants and animals. Further development of cloning technology relies on increased understanding of cell processes such as mitosis.

Section 17.2 Questions

1. Describe how nuclear transplants are used to clone frogs.
2. Dolly was not the first cloned animal, nor was she the first mammal clone. What made her cloning so special?
3. Explain why male animals would no longer be needed if cloning became the accepted method of reproduction.
4. If the nucleus is extracted from an adult animal cell and placed into an enucleated egg, how would it be possible to distinguish the cloned individual from the original?
5. Make a list of benefits and potential problems associated with cloning farm animals.
6. Speculate on the potential benefits and problems associated with cloning humans.
7. Research the nature versus nurture debate and the evidence provided by studies of twins. Find out about some psychological conditions that have both a genetic and an environmental component. What are the advantages and disadvantages of each approach? Think about the social, moral, and ethical implications of each viewpoint.

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17.3 Meiosis

meiosis two-stage cell division in which the chromosome number of the parental cell is reduced by half

haploid refers to the number of chromosomes in a gamete

diploid refers to twice the number of chromosomes in a gamete

homologous chromosomes paired chromosomes similar in shape, size, gene arrangement, and gene information

Meiosis is the type of cell division involved in the formation of sex cells, or gametes. In humans, this takes place in the testes and ovaries. Meiosis involves two stages of cell division that have some similarities to the phases in mitosis. In mitosis, the chromosome number of the daughter cells is the same as in the parent cell. In meiosis, the chromosome number of the daughter cells is half that of the parent cell. A human cell containing 46 chromosomes will undergo meiosis and produce gametes that have 23 chromosomes. Each gamete will contain both the same number and the same kind of chromosomes. The number of chromosomes in a gamete is called the **haploid** chromosome number, or n; the number of chromosomes in all other cells having a nucleus is twice the haploid number and is called the **diploid** number, or $2n$. In humans, the haploid chromosome number is 23 and the diploid chromosome number is 46.

Offspring carry genetic information from each of the parents. This explains why you might have your father's eyes but your mother's hair. Although you may look more like one parent than another, you receive genetic information from each parent. For example, your father gives you a chromosome with genes that code for eye colour, but so does your mother. Each of the 23 chromosomes that you receive from your biological father is matched by 23 chromosomes from your biological mother, so that each parent gives you half of your genetic information. The paired chromosomes are called **homologous chromosomes** because they are similar in shape, size, and gene arrangement (**Figure 1**). The genes in homologous chromosomes deal with the same traits. Each cell in your body, except the sex cells, contains 23 pairs of homologous chromosomes, or 46 chromosomes in total. The 23rd pair of chromosomes, which determine sex in mammals, are called the X and Y chromosomes and are only partially homologous. Males receive an X and a Y chromosome and females receive two X chromosomes. You will learn more about these chromosomes later in this chapter and in Chapter 22.

During fertilization, a haploid ($n = 23$) sperm cell unites with a haploid ($n = 23$) egg cell to produce a diploid ($2n = 46$) zygote. The fusion of male and female gametes restores the diploid chromosome number in the zygote. The zygote will begin dividing by mitosis and will eventually become a multicellular human baby.

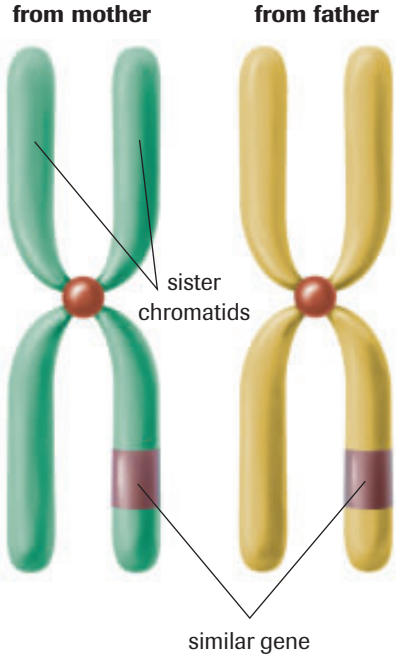


Figure 1
Homologous chromosomes

Stages of Meiosis

Meiosis involves two nuclear divisions that produce four haploid cells. Meiosis I is often called reduction division because the diploid, or $2n$, chromosome number is reduced to the haploid, or n, chromosome number. The second phase, meiosis II, is marked by a separation of the two chromatids. The phases used to describe the events of mitosis can also be used to describe meiosis. As with mitosis, DNA synthesis occurs prior to the cell division phase.

Meiosis I

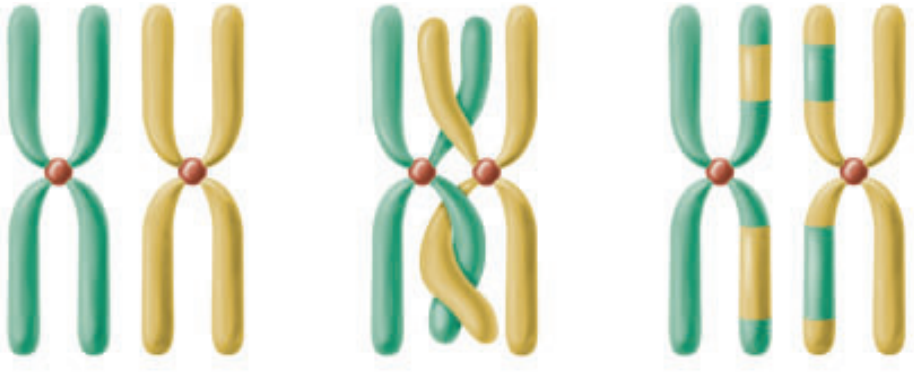
During prophase I, the nuclear membrane begins to dissolve, the centriole splits and its parts move to opposite poles within the cell, and spindle fibres are formed. The chromosomes come together in homologous pairs. Each chromosome of the pair is a homologue and is composed of a pair of sister chromatids. The whole structure is then referred to as a **tetrad** because each pair is composed of four chromatids.

tetrad a pair of homologous chromosomes, each with two chromatids

synapsis the pairing of homologous chromosomes

crossing over the exchange of genetic material between two homologous chromosomes

This process is referred to as **synapsis**. As the chromosomes synapse, the chromatids often intertwine. Sometimes the intertwined chromatids from different homologues break and exchange segments in a process called **crossing over** (**Figure 2**, next page). Crossing over permits the exchange of genetic material between homologous pairs of chromosomes.



homologous chromosome pair

As the chromosomes move closer together, synapsis occurs.

Chromatids break, and genetic information is exchanged.

Figure 2
Crossing over occurs between homologous pairs of chromosomes during prophase I of meiosis.

Metaphase I follows prophase I (**Figure 3**). The homologous chromosomes attach themselves to the spindle fibres and line up along the equatorial plate.

During anaphase I, the homologous chromosomes move toward opposite poles. The process is known as segregation. At this point of meiosis, reduction division occurs. One member of each homologous pair will be found in each of the new cells. Each chromosome consists of two sister chromatids.

During telophase I, a membrane begins to form around each nucleus. However, unlike in mitosis, the chromosomes in the two nuclei are not identical because each of the daughter nuclei contains one member of the homologous chromosome pair. Although homologous chromosomes are similar, they are not identical. They carry genes for the same traits (for example, eye colour), but those genes may differ (for example, coding for brown eyes or coding for blue eyes). The cells are now ready to begin the second stage of meiosis.

EXTENSION

Crossing Over

This Audio Clip will discuss the timing of crossing over and the benefit that a species derives from this process.

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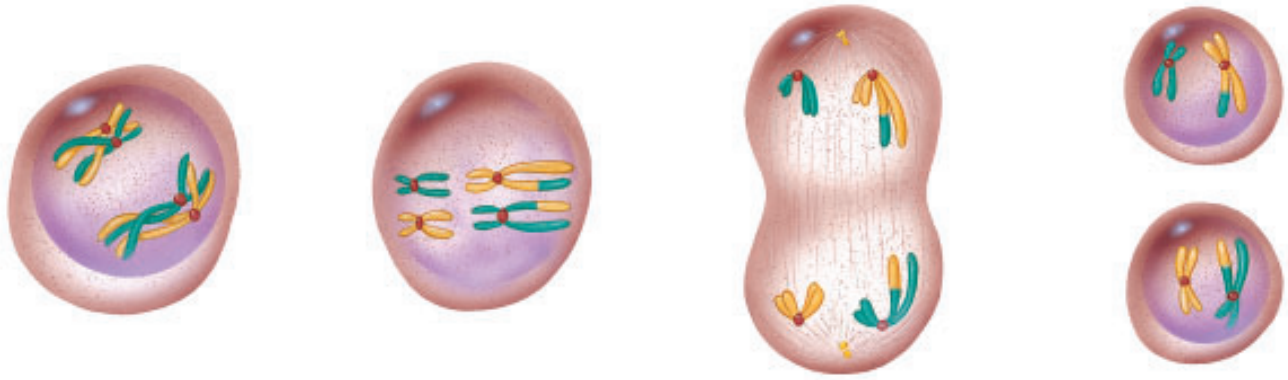


prophase I

The replicated chromosomes condense. Homologous chromosomes come together in synapsis and crossing over occurs. Chromosomes attach to the spindle.

metaphase I

Chromosomes line up at the equatorial plate.

anaphase I

Each chromosome separates from its homologue. They move to opposite poles of the cell.

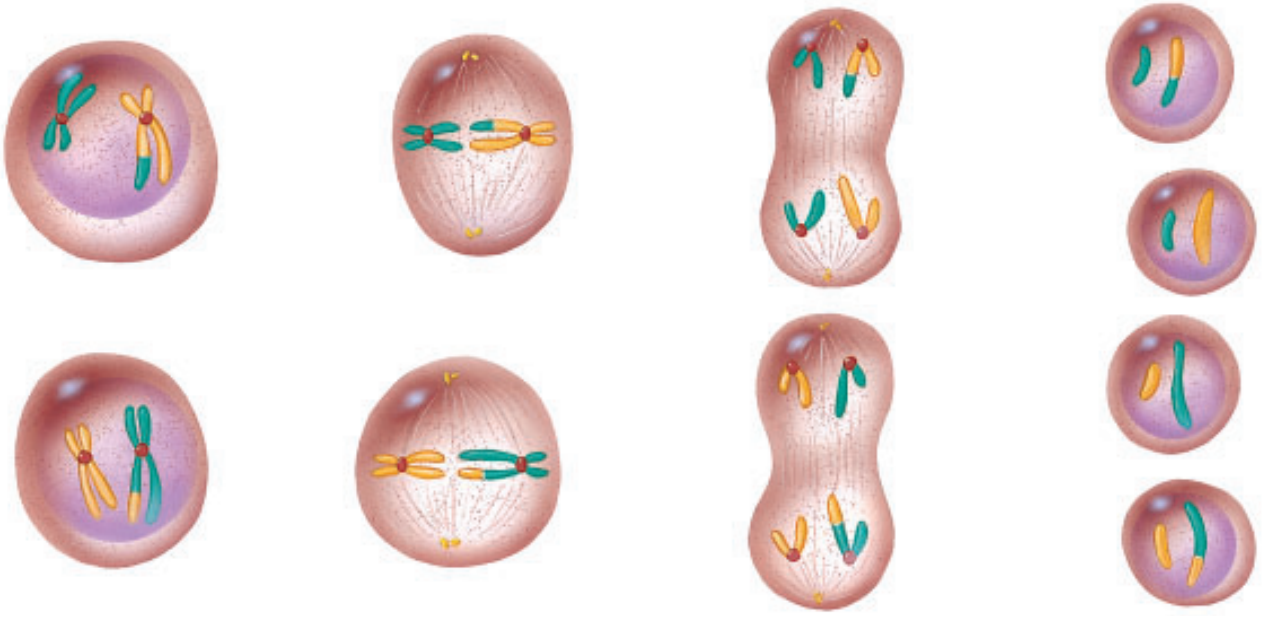
telophase I

The nucleus completes its division. The chromosomes are still composed of sister chromatids. The cytoplasm divides after telophase.

Figure 3
During meiosis I, homologous chromosomes are segregated.

Meiosis II

Meiosis II occurs at approximately the same time in each of the haploid daughter cells. However, for simplicity, consider the events in only one of the cells. (In **Figure 4**, both cells from meiosis I are shown). During meiosis II, pairs of chromatids will separate and move to opposite poles. Note that, unlike with mitosis and meiosis I, there is no replication of chromosomes prior to meiosis II.



prophase II

The centrioles in the two new cells move to opposite poles and new spindle fibres form. The chromosomes become attached to the spindle.

metaphase II

Chromosomes line up at the equatorial plate.

anaphase II

Sister chromatids of each chromosome separate and move to opposite poles.

telophase II

The cytoplasm separates, leaving four haploid daughter cells. The chromosome number has been reduced by half. These cells may become gametes.

Figure 4
During meiosis II, sister chromatids separate.

Prophase II signals the beginning of the second meiotic division. During this stage, the nuclear membrane dissolves and the spindle fibres begin to form.

Metaphase II follows prophase II. It is signalled by the arrangement of the chromosomes, each with two chromatids, along the equatorial plate. The chromatids remain pinned together by the centromere.

Anaphase II can be identified by the breaking of the attachment between the two chromatids and by their movement to the opposite poles. This stage ends when the nuclear membrane begins to form around the chromatids, now referred to as chromosomes.

The cell then enters its final stage of meiosis: telophase II. During this stage, the second nuclear division is completed and then the second division of cytoplasm occurs. Four haploid daughter cells are produced from each meiotic division.

Practice

1. Define meiosis. Describe the main stages in the process. Sketch the sequence of stages to help you in your description. Label your diagrams appropriately.
2. How are haploid cells different from diploid cells in humans?
3. What is a tetrad?
4. What are homologous chromosomes?
5. Do homologous chromosomes have the same number of genes? Explain.
6. Do homologous chromosomes have identical genes? Explain.

mini Investigation — Gamete Formation in Grasshoppers

Obtain prepared slides of grasshopper (**Figure 5**) testes and identify cells undergoing meiosis. Make a few sample diagrams of cells at various stages of cell division.

(a) Label the chromosomes.

(b) Are you able to count the chromosome number? Explain why or why not.

(c) Explain and compare what happens in prophase, metaphase, and anaphase of meiosis I and II.

(d) How do cells undergoing meiosis II differ from cells undergoing meiosis I?

Figure 5

Comparing Mitosis and Meiosis

Single-celled eukaryotic species undergo asexual reproduction by mitosis, followed by cytokinesis. In multicellular eukaryotic species, somatic cells undergo these same processes in order to grow and repair tissue. In contrast, meiosis occurs only in the sex cells of multicellular eukaryotic species, in order to produce the gametes needed for sexual reproduction.

The most significant difference between mitosis and meiosis is the end result (**Figure 6**). Mitosis results in two daughter cells that are identical to each other. The daughter cells have the same genetic information and carry the same number of chromosomes as the

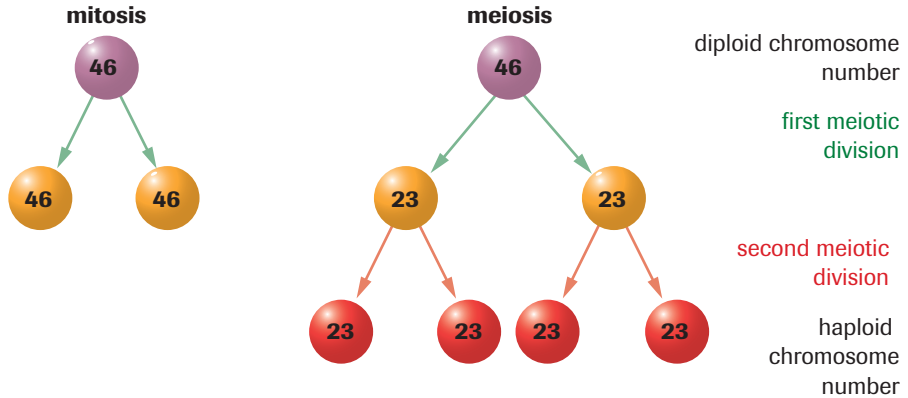
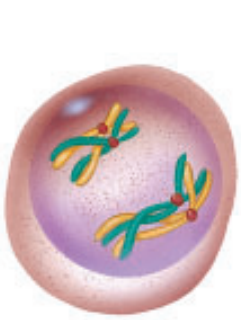


Figure 6
Comparison of mitosis and meiosis in humans. Mitosis produces two diploid cells from one diploid cell. Meiosis produces four haploid cells from one diploid cell.

parent cell. In contrast, meiosis results in four daughter cells that are different from each other and from the parent cell. The daughter cells have different genetic information from each other and from the parent cell and carry half the number of chromosomes as the parent cell.

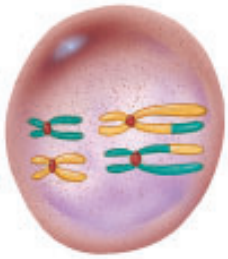
Figure 7 and **Figure 8** (next page) summarize the similarities and differences between mitosis and meiosis. As you examine **Figures 7** and **8**, make note of the chromosome number of the cell or cells, whether the chromosome number is haploid or diploid, and during which stage the chromosome number changes.

Meiosis, combined with fertilization, explains the variation in traits that is observed in species that reproduce sexually. The variation occurs through three mechanisms. First, crossing over during prophase I exchanges genes on the chromosomes. Second, during metaphase I, the paternal and maternal chromosomes are randomly assorted. Although homologues always go to opposite poles, a pole could receive all the maternal chromosomes, all the paternal ones, or some combination. Lastly, during fertilization, different combinations of chromosomes and genes occur when two gametes unite.



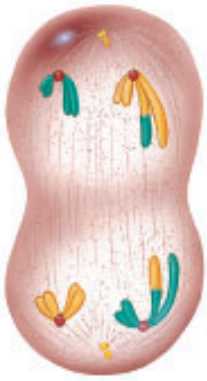
prophase I

The replicated chromosomes condense. Homologous chromosomes come together in synapsis and crossing over occurs. Chromosomes attach to the spindle.



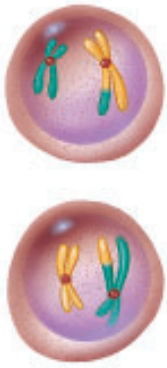
metaphase I

Homologous chromosomes line up at the equatorial plate.



anaphase I

Each chromosome separates from its homologue. They move to opposite poles of the cell.



telophase I

The nucleus completes its division. The chromosomes are still composed of sister chromatids. The cytoplasm divides after telophase.

Figure 7
Stages of meiosis I. During meiosis I, crossing over occurs and homologous pairs separate. These events do not occur during mitosis.

INVESTIGATION 17.3 ***Introduction***

Comparing Mitosis and Meiosis

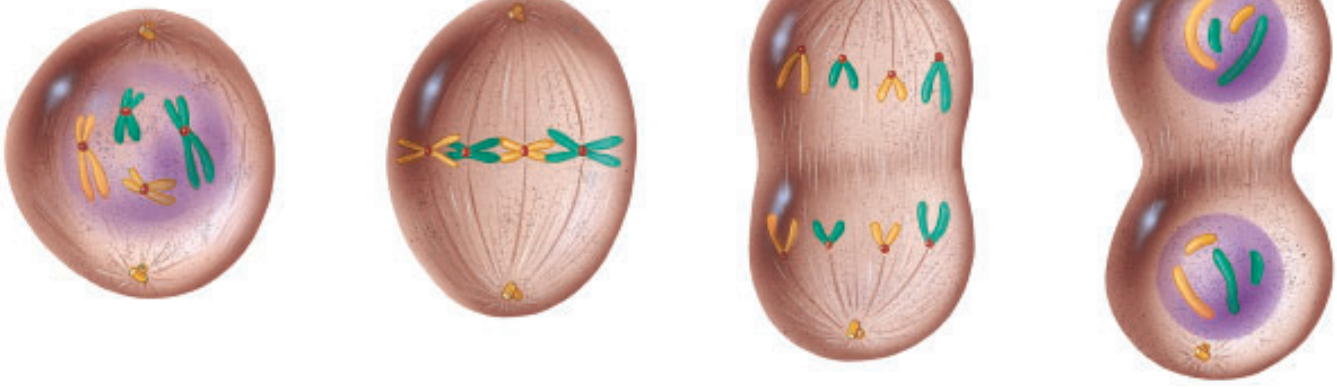
Scientists often use models to help them to understand complex processes. To understand the consequences of mitosis and meiosis, you must have a clear view of the similarities and differences between these two modes of cell division. In this investigation, you construct and use models to investigate these essential processes.

To perform this investigation, turn to page 590.

Report Checklist

● Purpose	○ Design	● Analysis
● Problem	○ Materials	● Evaluation
○ Hypothesis	○ Procedure	○ Synthesis
○ Prediction	○ Evidence	

(a) Mitosis



prophase

The chromosomes condense, becoming shorter and thicker. The centrioles assemble and spindle fibres attach to the centromeres of the chromosomes. The nuclear membrane starts to dissolve.

metaphase

Chromosomes line up at the equatorial plate. The nuclear membrane completely dissolves.

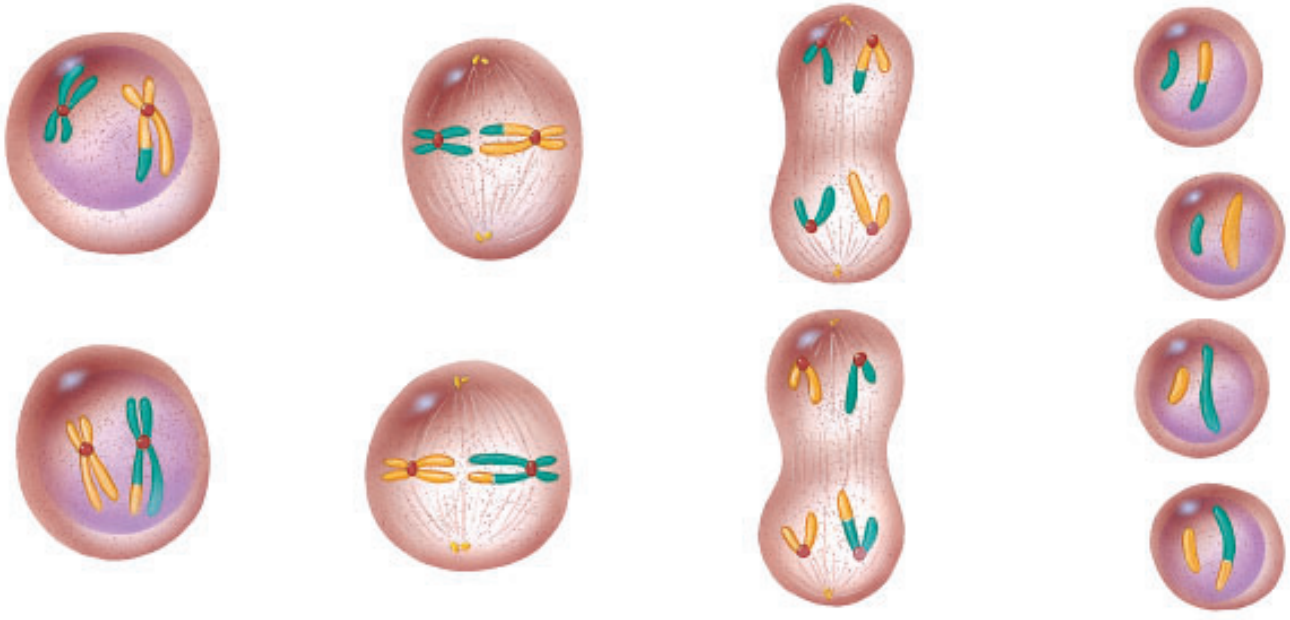
anaphase

The centromeres divide and the resulting chromosomes, formerly chromatids, move to opposite poles of the cell. An identical set of chromosomes moves to each pole.

telophase

Chromosomes lengthen again, the spindle fibres dissolve, and a nuclear membrane forms around the chromosomes.

(b) Meiosis II



prophase II

The centrioles in the two new cells move to opposite poles and new spindle fibres form. The chromosomes become attached to the spindle.

metaphase II

Chromosomes line up at the equatorial plate.

anaphase II

Sister chromatids of each chromosome separate and move to opposite poles.

telophase II

The cytoplasm separates, leaving four haploid daughter cells. The chromosome number has been reduced by half. These cells may become gametes.

Figure 8

Comparison of the stages in **(a)** mitosis and **(b)** meiosis II. In mitosis, homologous chromosomes are separated, giving rise to genetically identical sister cells. In meiosis II, the sister chromatids in the products of meiosis I separate as the cells divide again. This gives rise to four genetically different sex cells.

Practice

7. Copy and complete **Table 1**. Compare the chromosome number in the organisms before, during, and as a result of meiosis. Indicate whether the chromosome number is haploid or diploid.

Table 1 Chromosome Number in Cells of Four Organisms

	Human	Cat	Shrimp	Bean
Before meiosis				
chromosome number (haploid or diploid?)	46	?	?	?
number of pairs of homologous chromosomes	23	?	127	?
After meiosis I				
chromosome number (haploid or diploid?)	23	19	?	?
After meiosis II				
chromosome number (haploid or diploid?)	23	?	?	11
number of pairs of homologous chromosomes	0	?	?	?

Development of Male and Female Gametes

gametogenesis the formation of gametes (sex cells) in animals

ootid an unfertilized ovum

The formation of sex cells during meiosis is referred to as **gametogenesis**. Although human male and female gametes both follow the general process of meiosis, some differences do exist. The cytoplasm of the female gametes does not divide equally after each nuclear division. As shown in **Figure 9**, one of the daughter cells, called the **ootid**, receives most of the cytoplasm. The other cells, the polar bodies, die, and the nutrients are absorbed by the body of the organism. Only one ovum (egg cell) is produced from meiosis. In contrast, with sperm cells, there is an equal division of cytoplasm. Sperm cells have much less cytoplasm than egg cells. Sperm cells are specially designed for movement: they are streamlined and cannot carry excess weight. Egg cells use the nutri-

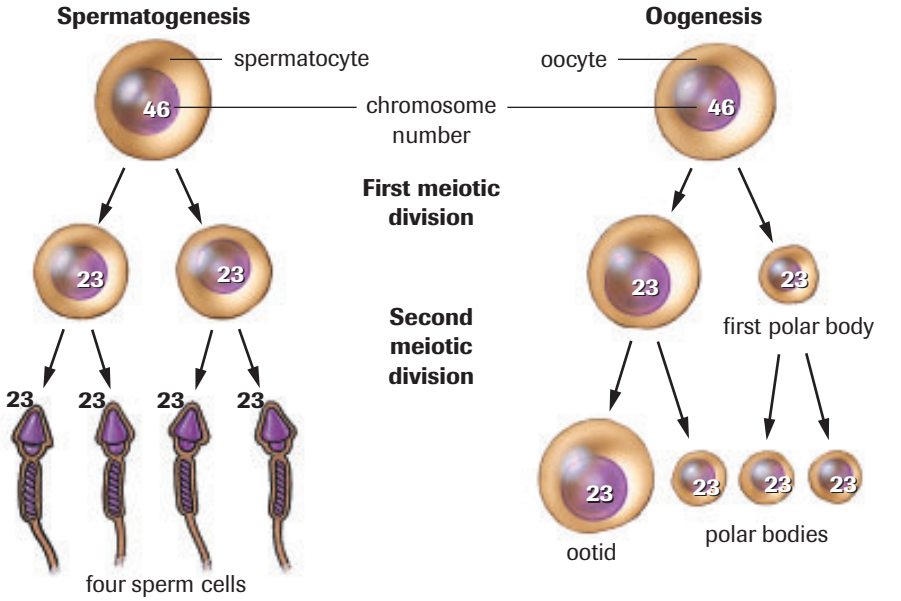


Figure 9
Generalized diagram of sperm and egg cell formation in humans

ents and organelles carried within the cytoplasm to fuel future cell divisions in the event that the egg cell becomes fertilized.

Human males make many more sex cells than females. The diploid spermatocytes—the cells that give rise to sperm cells—are capable of many mitotic divisions before meiosis ever begins. Males can produce one billion sperm cells every day. At birth, human females have about two million primary oocytes in their ovaries. Primary oocytes have already entered meiosis I, but they will remain suspended in prophase I until the female reaches reproductive age, or puberty. Starting at the first menstrual cycle, meiosis will resume in one oocyte at a time, once a month.

WEB Activity

Case Study—Comparing Life Cycles of Plants

In this Web-based Case Study, you will observe and compare the life cycles of different plants. By examining the reproductive life cycles of plants you will gain a greater understanding of how reproductive diversity contributes to the evolution of complex organisms.

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Cell Division and Life Cycles

Organisms that undergo asexual reproduction produce offspring by mitosis. In this type of life cycle, cells divide by mitosis and give rise to daughter cells with the same chromosome number as the parent cell. There is no change in chromosome number. Examples of organisms that reproduce asexually are bacteria and yeasts.

In contrast, the chromosome number changes during the life cycle of a species that undergoes sexual reproduction. Examples of sexually reproducing species include flowering plants and birds. Two events in sexual reproduction change chromosome number: meiosis and fertilization. The gametes are formed by meiosis; these cells have half the chromosome number as the somatic cells. During fertilization, two gametes join to form a zygote, and the chromosome number is restored to that of the somatic cells.

There are variations in these two main types of the life cycles. **Figure 10**,on the next page shows a common life cycle found in flowering plants. In flowering plants, pollen contains the male sex cells, and the female egg cells are stored within the flower. The gametes contain a haploid chromosome number ($1n$). At fertilization, a diploid zygote ($2n$) is formed. The zygote undergoes mitosis to produce seeds, which then undergoes further mitosis to produce the adult $2n$ plant, called the sporophyte. Specialized cells in the mature $2n$ plant undergo meiosis to produce haploid ($1n$) spores. The spores then undergo mitosis to produce a mature, multicellular gametophyte. In most flowering plants, the gametophyte is too small to see without magnification. Since mitosis does not change chromosome number, the gametophyte is also haploid ($1n$). Specialized cells in the gametophyte develop into gametes, and the cycle begins again. Many familiar plants are sporophytes, such as the pine trees in a boreal forest. In other plant species, such as ferns, it is the gametophyte that is the larger, dominant form.

Figure 11,on the next page shows a common life cycle for animals, such as humans. In this life cycle, the gametes (sperm cells and egg cells) are haploid ($1n$) and single-celled. During fertilization, the gametes fuse and form a diploid ($2n$) zygote. This zygote undergoes mitosis to form the multi-cellular diploid adult body. Specialized cells in the adult body (in humans, cells in the testes and ovaries) undergo meiosis to produce gametes. Up to this point, the life cycles of plants in **Figure 10** and of animals in **Figure 11** are the same. However, the gametes of most animals do not undergo mitosis to form a multi-cellular gametophyte. Instead, the haploid stage remains single celled. When these haploid gametes unite, fertilization occurs and the life cycle begins again.

EXTENSION

Reproductive Strategies for Survival (Non-Human)

The different species on our planet have a remarkable variety of strategies to ensure their survival. Review some of these reproductive strategies by completing this extension activity.

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DID YOU KNOW?

Two Styles of Life Cycle

Some species undergo both sexual and asexual life cycles. For example, the spider plant can reproduce by seeds (sexual reproduction) or by runners (asexual reproduction). Aphid females reproduce asexually when the environment is stable, and sexually when the environment changes. Similarly, the male drones in a honey bee colony are produced by asexual reproduction, but the female workers and the queens are products of sexual reproduction.

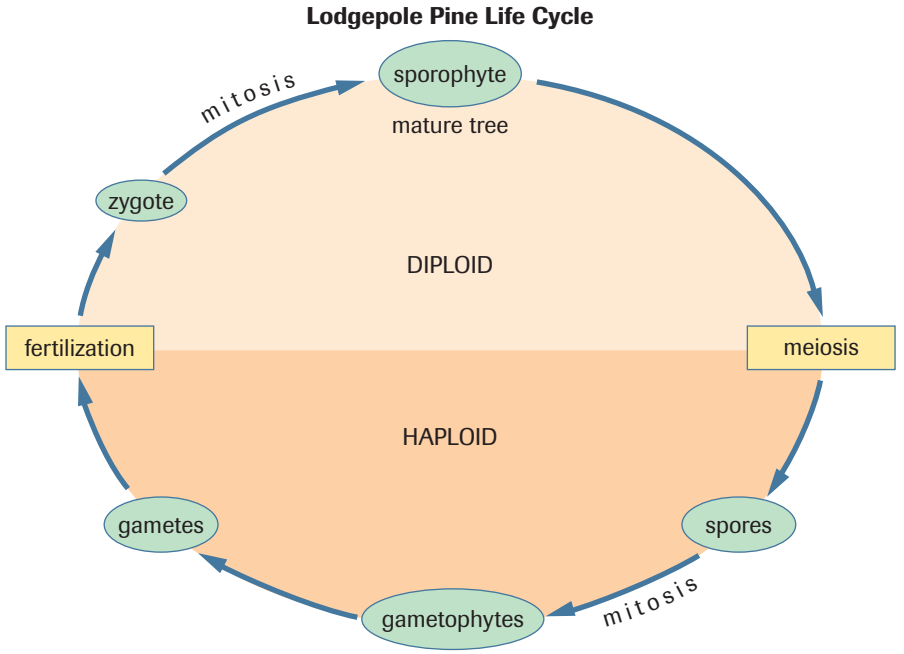

Figure 10
Lodgepole pine life cycle. The diploid cells formed at fertilization undergo mitosis to form the multicelled *sporophyte (the tree)*. The haploid stage starts when meiosis produces spores. These undergo mitosis to form a multicellular gametophyte, which is contained in the cones.

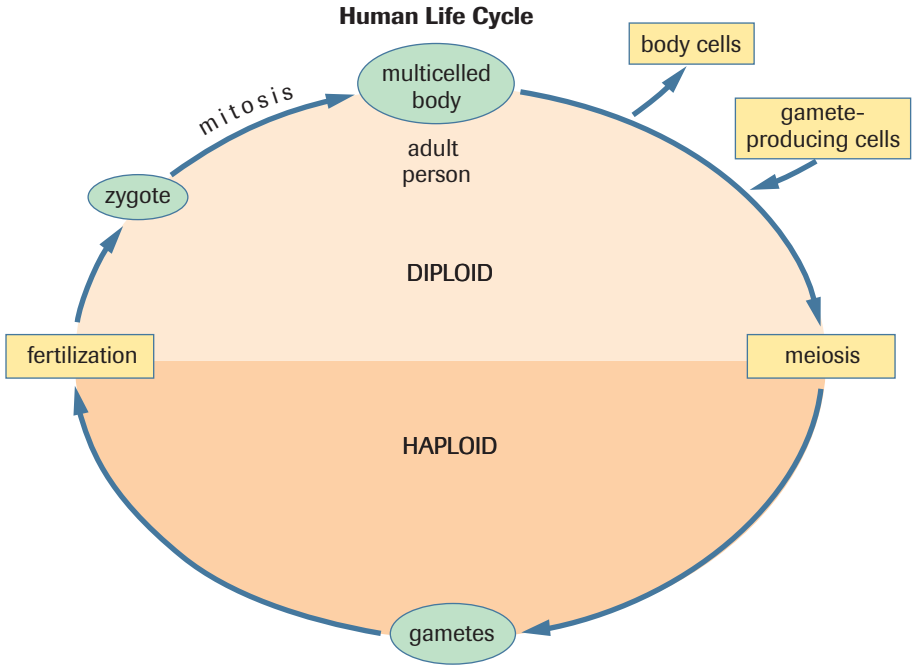

Figure 11
Human life cycle. The diploid cells formed at fertilization undergo mitosis to form the multicelled body. The haploid stage is the single-celled gametes.

SUMMARY Meiosis

- Meiosis involves the formation of sex cells or gametes. All gametes produced by meiosis have haploid chromosome numbers.
- Cells undergoing meiosis pass through two divisions.
- Homologous chromosomes are similar in shape, size, gene arrangement, and gene information.
- Crossing over is the exchange of genetic material between homologous chromosomes that occurs during meiosis.

Section 17.3 Questions

1. How does the first meiotic division differ from the second meiotic division?
2. Explain why synapsis may lead to the exchange of genetic information.
3. Construct a table to compare meiosis with mitosis. How does meiosis differ from mitosis?
4. A muscle cell of a mouse contains 22 chromosomes. Based on this information, how many chromosomes are there in the following types of mouse cells?
   (a) daughter muscle cell formed from mitosis
   (b) egg cell
   (c) fertilized egg cell
5. Compare the mechanisms of gametogenesis in males and females.
6. When meiosis occurs in females, the cytoplasm is not divided equally among the resulting four cells. Explain why.
7. Compare the life cycles of plants and animals.
8. **Figure 12** shows sperm cell production following meiosis.
   (a) Which cells do not contain homologous pairs?
   (b) If the chromosome number for cell A is 12, indicate the chromosome number for cell C.

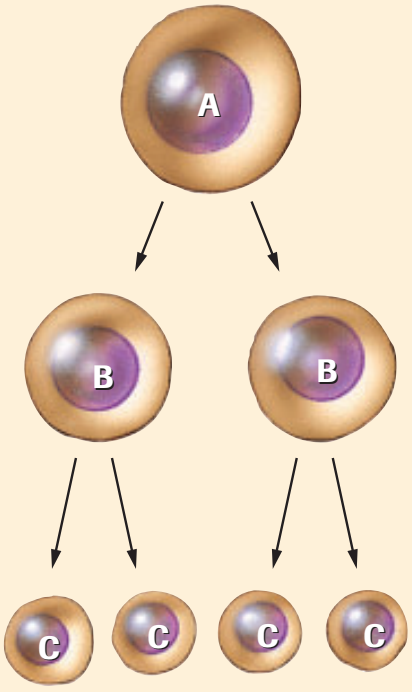

Figure 12
Sperm cell production in humans

9. Use **Figure 13** to answer the questions below.
   (a) Which process(es) identify mitosis? Explain your answer.
   (b) Which process(es) identify meiosis? Explain your answer.

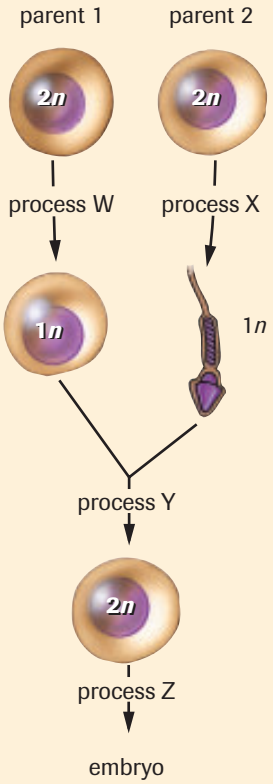

Figure 13
The processes and number of sets of chromosomes involved in the production of an embryo in humans

10. King Henry VIII of England had some of his wives executed for not producing sons. Indicate why a little knowledge of meiosis might have been important for Henry's wives.
11. A microscopic water animal called *Daphnia* can be reproduced from an unfertilized egg. This form of reproduction is asexual because male gametes are not required. Indicate the sex of the offspring produced. Explain your answer.

17.4 Abnormal Meiosis

nondisjunction the failure of a pair of homologous chromosomes to separate properly during meiosis

Meiosis, like most processes of the body, is not immune to mistakes. **Nondisjunction** occurs when two homologous chromosomes fail to separate during meiosis or mitosis. The result is that one of the daughter cells will have too many chromosomes, while the other will have too few. Cells that lack genetic information, or have too much information, will not function properly. Nondisjunction can also occur in any cell during mitosis, but the effects are most devastating during the formation of sex cells in meiosis.

polyploidy a condition in which an organism has more than two complete sets of chromosomes

Some organisms have more than two complete chromosome sets. This condition is called **polyploidy**. Polyploid organisms may have three chromosome sets (triploidy or $3n$), four chromosome sets (tetraploidy or $4n$), and rarely, even more than four chromosome sets. Polyploidy can result when a diploid ($2n$) egg cell is fertilized by a haploid ($1n$) sperm, giving rise to a $3n$ cell. Nondisjunction of all chromosomes within the egg cell produces a diploid sex cell, which then becomes triploid upon fertilization. Tetraploid organisms are most often produced by the failure of the $2n$ zygote to divide after replicating its chromosomes. Following normal mitosis a $4n$ embryo is formed. Polyploidy is common in plants. Wheat, oats, tobacco, and potatoes are agriculturally important polyploid species. Plant geneticists may use chemicals that create errors in meiosis and mitosis to create new polyploid plants.

trisomy the condition in which there are three homologous chromosomes in place of a homologous pair

monosomy the condition in which there is a single chromosome in place of a homologous pair

In humans, nondisjunction produces gametes with 22 and 24 chromosomes. The gamete with 24 chromosomes has both chromosomes from one of the homologous pairs. If that gamete joins with a normal gamete of 23 chromosomes from the opposite sex, a zygote containing 47, rather than 46, chromosomes will be produced. The zygote will then have three chromosomes in place of the normal pair. This condition is referred to as **trisomy**. However, if the sex cell containing 22 chromosomes joins with a normal gamete, the resulting zygote will have 45 chromosomes. The zygote will have only one of the chromosomes rather than the homologous pair. This condition is called **monosomy**. Once the cells of the trisomic or monosomic zygotes begin to divide, each cell of the body will contain more or fewer than 46 chromosomes.

Figure 1
Dr. Renée Martin

WEB Activity

Canadian Achievers—Dr. Renée Martin

Pregnancy loss, birth defects, and mental retardation have been linked with chromosome abnormalities in sperm and eggs, but much of the scientific research to date has focused on abnormalities in the egg. Dr. Renée Martin (**Figure 1**), a medical geneticist from the University of Calgary, is recognized for her research on chromosomal abnormalities in human sperm cells. A research centre at the university has been named after her. Dr. Martin's research indicates that 10 % of sperm in normal men have a chromosomal abnormality, but men who have undergone radiotherapy have much higher frequencies of abnormal sperm. One of the most important questions to be answered is whether or not any of these abnormal sperm cells actually fertilize an egg. Dr. Martin's research will provide valuable information on birth defects and miscarriages. Visit the Nelson Web site to learn more about Dr. Martin's research contributions.

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Nondisjunction Disorders

Nondisjunction is associated with many different human genetic disorders. For example, Down syndrome is a trisomic condition. Down syndrome is also called trisomy 21 because it usually results from three copies of chromosome 21. People with Down syndrome (**Figure 2**) can be identified by several common traits, regardless of race: a round, full face; enlarged and creased tongue; short height; and a large forehead. Down syndrome is generally associated with mental retardation, although people with this condition retain a wide range of mental abilities. The risk of having a baby with Down syndrome increases with the age of the mother. About 1 in 600 babies is born with Down syndrome.

Turner syndrome occurs when sex chromosomes undergo nondisjunction. This monosomic disorder produces a female with a single X chromosome. In the egg cell, both homologous X chromosomes move to the same pole during meiosis I (**Figure 3**). When the egg with no X chromosome is fertilized by a normal sperm cell with an X chromosome, a zygote with 45 chromosomes is produced. Individuals with Turner syndrome appear female, but do not usually develop sexually and tend to be short and have thick, widened necks. About 1 in every 3000 female babies is a Turner syndrome baby. Most Turner syndrome fetuses are miscarried before the 20th week of pregnancy.

Klinefelter syndrome is caused by nondisjunction in either the sperm or egg (**Figure 3**). The child inherits two X chromosomes—characteristic of females—and a single Y chromosome—characteristic of males. The child appears to be a male at birth; however, as he enters sexual maturity, he begins producing high levels of female sex hormones. Males with Klinefelter syndrome are sterile. It has been estimated that Klinefelter syndrome occurs, on average, in 1 of every 500 male babies.

Figure 2
People with Down syndrome have a wide range of abilities.

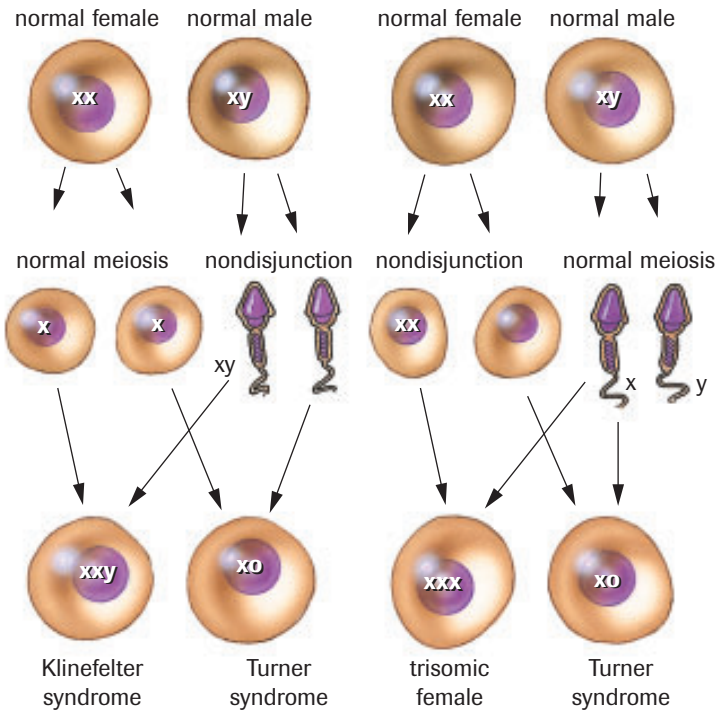

Figure 3
Nondisjunction disorders in humans

CAREER CONNECTION

Geneticist

Geneticists are professionals with specialized education, training, and experience in genetics. Those with expertise in medical genetics may help families understand birth defects and how diseases are inherited. They may counsel people who carry genes that increase their risk of developing disease, such as some forms of cancer.

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Karyotype Charts

karyotype chart a picture of chromosomes arranged in homologous pairs

One tool for detecting the results of abnormal meiosis is a chart of the chromosomes called a karyotype. Technicians obtain a **karyotype chart** by mixing a small sample of tissue with a solution that stimulates mitotic division. A different solution is added which stops division at metaphase. Since chromosomes are in their most condensed form during metaphase—their size, length, and centromere location are most discernible—it is the best phase in which to obtain a karyotype. The metaphase cells are placed onto a slide and then stained, so that distinctive bands appear. A photograph of the chromosomes is taken. The image is enlarged, and each chromosome is cut out and paired up with its homologue. Homologous chromosomes are similar in size, length, centromere location, and banding pattern. Finally, all the pairs are aligned at their centromeres in decreasing size order. The sex chromosomes are always placed last.

Figure 4 shows karyotypes of a normal male and of a female with Down syndrome. In about 95 % of cases, a child with Down syndrome has an extra chromosome in chromosome number 21. This trisomic disorder is produced by nondisjunction; the person has too much genetic information. Compare the chromosomes of a male shown in **Figure 4** (**a**), with the chromosomes of a female who has Down syndrome, shown in **Figure 4** (**b**). Notice how the chromosomes are arranged in pairs.

EXTENSION

Karyotype Preparation

This animation depicts the steps involved in preparing a karyotype chart. You can also see representative karyotypes from individuals with nondisjunction disorders.

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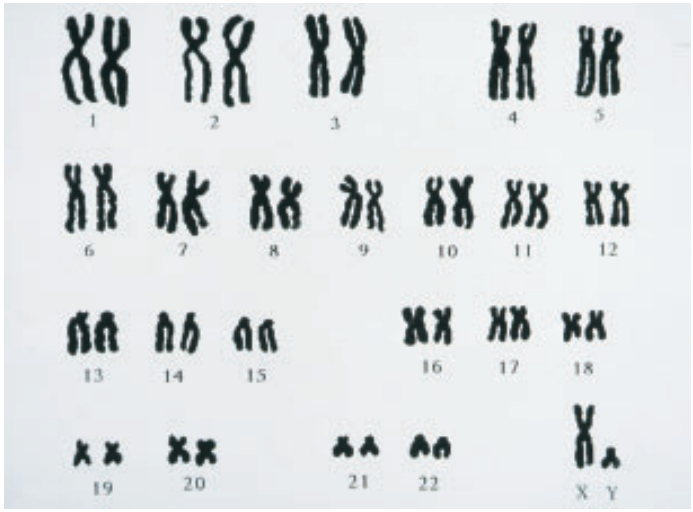
(a)



(b)

Figure 4

(a) Karyotype chart of a male with 46 chromosomes. Notice that the chromosome pair number 23 is not homologous. Males contain an X and a Y chromosome. They act as a homologous pair in meisois, but they are not similar in size and shape as are the other chromosome pairs.

(b) Karyotype of a female with Down syndrome. Note the trisomy of number 21. Down syndrome affects both males and females.

SAMPLE exercise 1

Figure 5 shows the incomplete karyotype chart of a human. Notice that several chromosomes are missing. Identify where chromosomes a to f (**Figure 6**) should be in this karyotype chart.

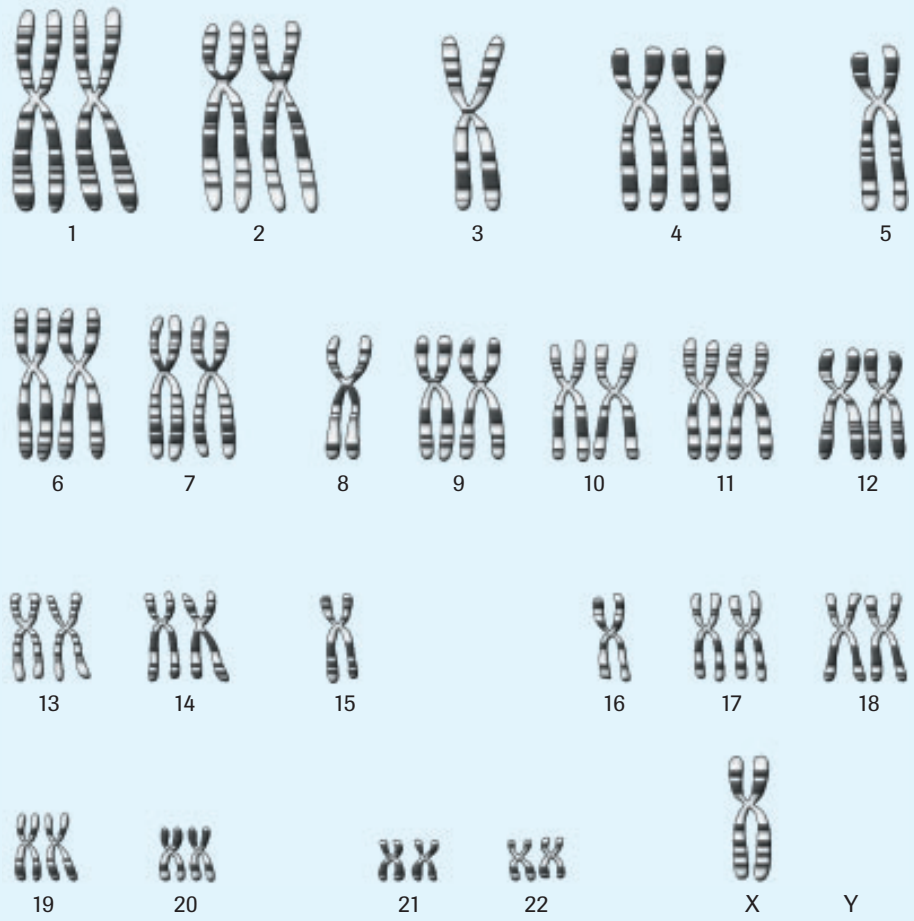


Figure 5

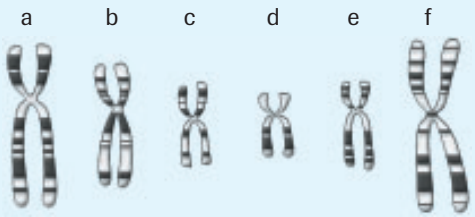


Figure 6

Solution

1. Start by scanning the karyotype chart to see which pairs are missing a chromosome. Pairs 3, 5, 8, 15, and 16 need a partner.
2. Match the most obvious chromosomes first: the longest, shortest, or most distinctively banded chromosomes.
3. For chromosome matches that are not as obvious, look carefully at the banding pattern and location of the centromere.

DID YOU KNOW?

Amniocentesis

A diagnostic technique known as amniocentesis can be used to test for nondisjunction and other genetic disorders in developing fetuses. During this procedure, a fine needle is inserted into the amniotic sac that surrounds the fetus, and about 10 mL of the amniotic fluid in which the fetus is bathed is withdrawn. This fluid contains fetal cells that can be used to produce a karyotype chart, as well as chemicals that may signal specific disorders.

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Learning Tip

You can also construct a karyotype chart using a copy of the chromosome images. For the Sample Exercise and Practice question 1, copy **Figures 5**, **6** and **7**. Then, cut out the chromosome images in **Figures 6** and **7**, and position them on **Figure 5** according to their size, shape, and banding patterns.

4. Always pay attention to the X and Y chromosomes. In **Figure 5**, on the previous page, the missing chromosome might be X or Y. If it is Y, it will have to be found through elimination since it will not match X.

a, 5 b, 8 c, 16 d, Y e, 15 f, 3

Practice

1. This person has either Down syndrome or Klinefelter syndrome. Identify the placement of chromosome g (**Figure 7**) to identify which of these two disorders the patient has.

g

Figure 7

EXTENSION

Karyotyping

There are a number of human genetic disorders that involve nondisjunction. In this Virtual Biology Lab, you will construct karyotype charts and use them to predict genetic disorders, in much the same way as a genetic counsellor might.

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WEB Activity

Web Quest—Modelling Mitosis and Meiosis

Cellular division is one of the most critical processes an organism regularly undergoes. Unfortunately, errors during cellular division can result in a number of genetic syndromes such as Down syndrome, Turner syndrome, Klinefelter syndrome, and XYY syndrome. In this Web Quest, you will explore normal and abnormal cellular division. You will use the knowledge that you gathered to create an animation or presentation that shows exactly how abnormal cellular divisions occur.

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SUMMARY Abnormal Meiosis

- Nondisjunction occurs when two homologous chromosomes move to the same pole during meiosis. In humans, this produces gametes with 22 and 24 chromosomes.
  - Trisomy: a zygote containing 47 chromosomes; causes human genetic disorders such as Down syndrome and Klinefelter syndrome
  - Monosomy: a zygote containing 45 chromosomes; causes Turner syndrome
- A karyotype chart is a picture of chromosomes arranged in homologous pairs in descending order by size, with the sex chromosomes placed last.

Section 17.4 Questions

1. What is nondisjunction?

2. Differentiate between monosomy and trisomy.

3. What is Down syndrome?

4. What is a karyotype?

5. What is Turner syndrome?

6. Use a diagram to illustrate how nondisjunction in meiosis I ($2n = 4$) differs from nondisjunction in meiosis II.

INVESTIGATION 17.1

Frequency of Cell Division

Report Checklist

● Purpose	○ Design	● Analysis
● Problem	○ Materials	● Evaluation
○ Hypothesis	○ Procedure	● Synthesis
○ Prediction	● Evidence	

In this activity, you will view and compare cells from onion cells and from a whitefish blastula in various stages of mitosis. Because slides are used, the cell divisions you will be viewing are frozen in time. Therefore, it will not be possible for you to watch a single cell progress through the stages of mitosis. Based on your observations, you will determine the frequency of cell division and construct a clock representing the division cycle, given the time taken to complete one cycle of mitosis. In a table, you will record the number of cells in each stage of mitosis.

Materials

microscope
lens paper
prepared slides of onion root tip
prepared slides of whitefish blastula

Procedure

Part 1: Observing Dividing Cells

1. Obtain an onion root tip slide and place it on the stage of your microscope. View the slide under low-power magnification. Focus using the coarse-adjustment knob.
2. Centre the root tip in the field of view and then rotate the nosepiece to the medium-power objective lens. Focus the image using the fine-adjustment knob. Observe the cells near the root cap. This area is referred to as the meristematic region of the root.
3. Move the slide to view the cells away from the root tip. These are the mature cells of the root. Record the differences between the cells of the meristematic area and the mature cells of the root. Draw a diagram to help you (**Figure 1**).
4. Return the slide to the meristematic area and centre the root tip. Rotate the nosepiece to the high-power objective lens. Use the fine adjustment to focus the image.
5. Locate and observe cells in each of the phases of mitosis. It will be necessary to move the slide to find each of the four phases. Use **Figure 1** as a guide. Draw, label, and title each of the phases of mitosis. It is important to draw only the structures that you can actually see under the microscope.

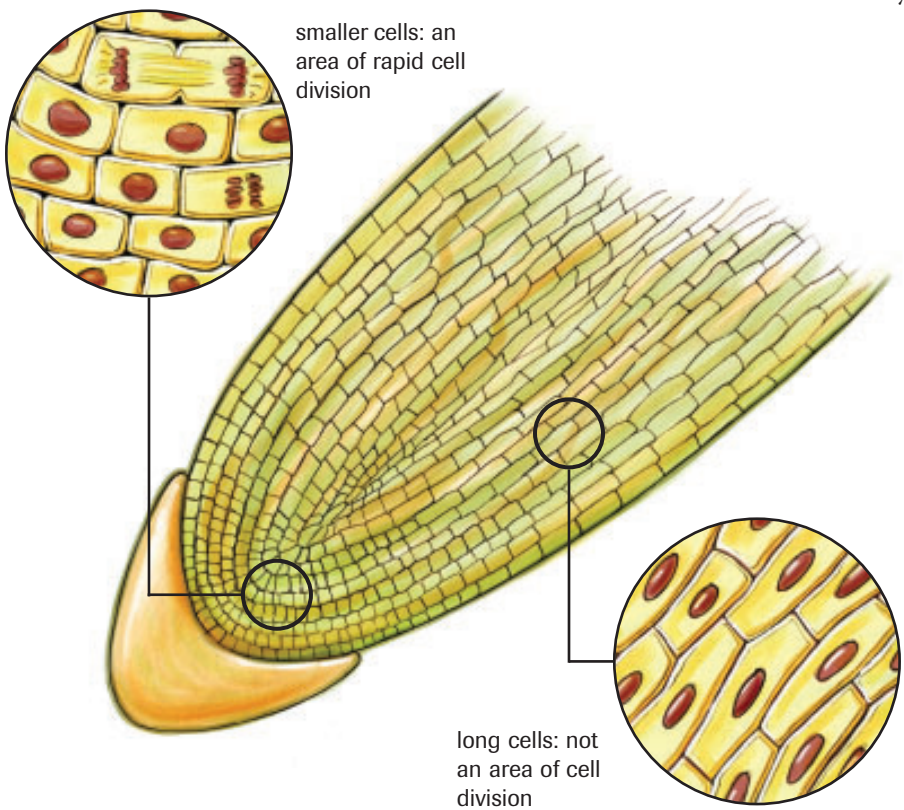

Figure 1
Meristematic region of the onion root tip where the cells are actively growing and dividing

6. Return your microscope to the low-power objective lens and remove the slide of the onion. Place the slide of the whitefish blastula on the stage. Focus with the coarse-adjustment knob. Repeat the procedure that you followed for the onion cells and, in the whitefish blastula, locate dividing cells under high-power magnification. Note how different the animal cells are compared to the plant cells.

Part 2: Determining the Frequency of Cell Division

7. Count 20 adjacent whitefish blastula cells and record whether the cells are in interphase or division phase. Record the number of cells in interphase and the number of cells that are actively dividing.
8. Repeat the same procedure for the meristematic region of the plant root.

Part 3: Creating a Cell-Division Clock

9. Under high-power magnification, locate 50 onion root cells that are dividing. Do not include cells that are between divisions. Identify the phase of mitosis each cell is in. Record the number of cells in each phase.
10. Repeat the procedure for the cells of the whitefish blastula.

Analysis and Evaluation

Part 1: Observing Dividing Cells

(a) How do the cells of the meristematic area differ from the mature cells of the root?

(b) Why were plant root tip cells and animal blastula cells used for viewing cell division?

(c) Explain why the cells that you viewed under the microscope do not continue to divide.

(d) Compare and contrast cell division in plant and animal cells. Use a Venn diagram to organize your ideas.

Part 2: Determining the Frequency of Cell Division

(e) For both the plant and animal cells, calculate the percentage of cells that are dividing. Use the following formula:

$$\frac{\text{Number of cells dividing}}{\text{Total number of cells counted}} \times 100 = \_\_\_ \% \text{ dividing}$$

(f) For both plant and animal cells, create a circle graph showing the percentage of cells in division phase and the percentage of cells in interphase. Label the diagrams appropriately. Compare the graphs. How are they different? How are they the same?

Part 3: Creating a Cell-Division Clock

(g) For both plant and animal cells, calculate the percentage of cells that are in each of these four phases: prophase, metaphase, anaphase, and telophase.

(h) For each cell type, construct a circle graph showing the percentage of cells in each phase of mitosis. Include labels and titles.

(i) If it takes 16 h to complete one cycle of mitosis for whitefish and 12 h for onions, determine the time spent in each phase. Include this information in your circle graphs.

Synthesis

(j) The number of animal cells in each phase of mitosis was recorded in **Table 1**. If the time taken to complete one cycle of mitosis was 15 h, create a cell-division clock to represent the data.

Table 1 Number of Cells in Different Phases of Mitosis

Mitotic phase	Number of cells in phase
prophase	15
metaphase	20
anaphase	10
telophase	5

INVESTIGATION 17.2

Identification of a Cancer Cell

Report Checklist

○ Purpose	● Design	● Analysis
● Problem	○ Materials	● Evaluation
○ Hypothesis	○ Procedure	● Synthesis
○ Prediction	● Evidence	

Purpose

To identify cancerous cells and to recognize the differences between cancerous and non-cancerous cells

Materials

light microscope
lens paper
prepared slide of squamous cell carcinoma

Procedure

1. Clean the microscope lenses with lens paper. Rotate the revolving nosepiece to the low-power objective lens. Place the slide of the carcinoma on the stage of the microscope and bring the image into focus using the coarse-adjustment knob.
2. Locate the dermal and epidermal layers. Draw a line diagram showing the position of the epidermal and dermal cell layers. Determine and record whether the cells of the epidermis are invading the dermis.
3. Rotate the revolving nosepiece to the medium-power objective lens. Locate a cancerous cell. **Figure 1** is an example of cancerous cells. Use the fine-adjustment knob to bring the image into focus. Observe how cells of the carcinomas have a much larger nucleus. They appear pink in colour and often have an irregular shape.

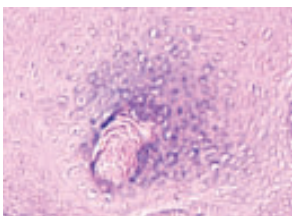

Figure 1

4. Rotate the nosepiece to high-power magnification, and bring the image into focus using the fine-adjustment knob.
5. Estimate and record the size of the cell, in micrometres (μm).
6. Estimate and record the size of the nucleus of the same cell, in micrometres (μm).
7. Rotate the revolving nosepiece to the medium-power objective lens and locate a normal cell. Rotate the nosepiece to the high-power objective lens, and bring it into focus with the fine-adjustment knob. **Figure 2** is an example of normal cells.

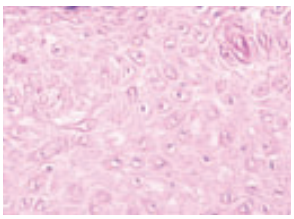

Figure 2

8. Repeat steps 5 and 6 for the normal cell.

Analysis

(a) Using the formula below, determine the nucleus-to-cytoplasm ratio for the cancerous cell and for the normal cell.

Evaluation

(b) Compare the cancerous and normal cells in a table similar to **Table 1**.

Table 1

Cell type	Cell size	Nuclear shape	Nuclear size	Nucleus-to-cytoplasm ratio
normal cell				
cancerous cell				

INVESTIGATION 17.2 *continued*

Synthesis

(c) Cancerous cells are often characterized by a large nucleus. Based on what you know about cancer and cell division, provide an explanation for the enlarged nucleus.

(d) Why are malignant (cancerous) tumors a greater threat to life than benign tumors?

(e) Provide a hypothesis that explains why the skin is so susceptible to cancer.

(f) A scientist finds a group of irregularly shaped cells in an organism. The cells demonstrate little differentiation, but the nuclei in some of the cells stain darker than others.

(i) Based on these findings, would it be logical to conclude that the organism has cancer? Justify your answer.

(ii) What additional tests might be required to prove or disprove the hypothesis that the cells are cancerous?

INVESTIGATION 17.3

Report Checklist

● Purpose	○ Design	● Analysis
○ Problem	○ Materials	● Evaluation
○ Hypothesis	○ Procedure	○ Synthesis
○ Prediction	○ Evidence	

Comparing Mitosis and Meiosis

In this investigation, you will model and compare the events of mitosis and meiosis. In this model, you will create homologous chromosomes that have the same size and shape, but different colours. This will show that they are similar but not identical.

Materials

red modelling clay
blue modelling clay
green modelling clay
plastic knife
sheets of paper
pencil

Procedure

For each step, make a coloured sketch of your model with appropriate labels. Include brief descriptions of your steps and make sure to use the same step numbers as given.

Part 1: Mitosis

1. Take some red clay and roll it between your hands to create a piece 10 cm long and about as thick as your finger. Make another piece about 5 cm long.
2. Repeat step 1 with the blue clay.
3. Make an identical copy of each piece of clay. Then attach the identical pieces with a green ball of clay (**Figure 1**).
4. Draw a line down the length of a sheet of paper. Line up the four chromosomes along the line (**Figure 2**).

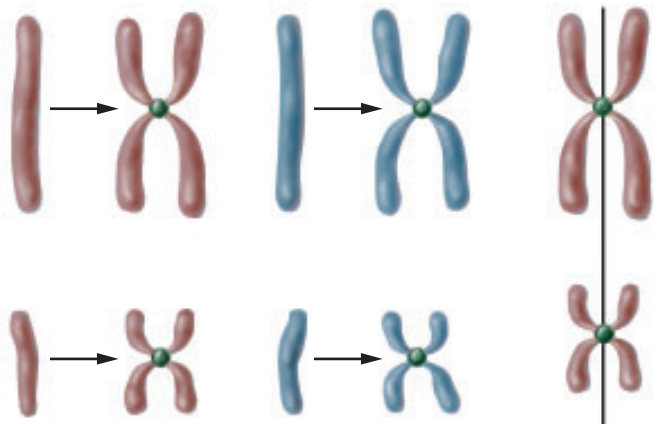

Figure 1

5. Remove the green balls and move each of the single pieces of clay to opposite ends of the paper (**Figure 3**, next page).
6. Before every mitotic division, each chromosome is duplicated during interphase. Make an identical copy of each piece of clay as before (**Figure 4**, next page).

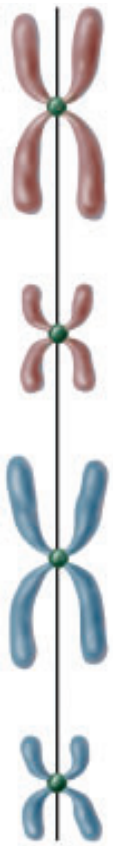

Figure 2

Part 2: Meiosis

7. Follow steps 1 to 3 from part 1.

INVESTIGATION 17.3 *continued*

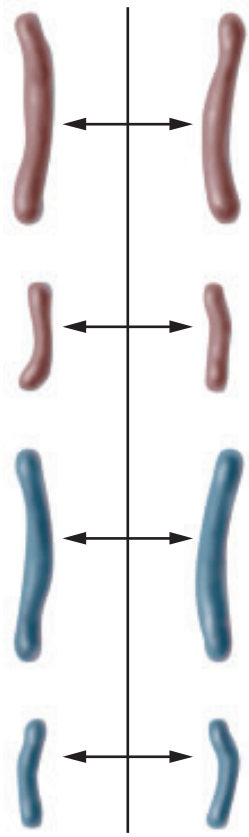

Figure 3

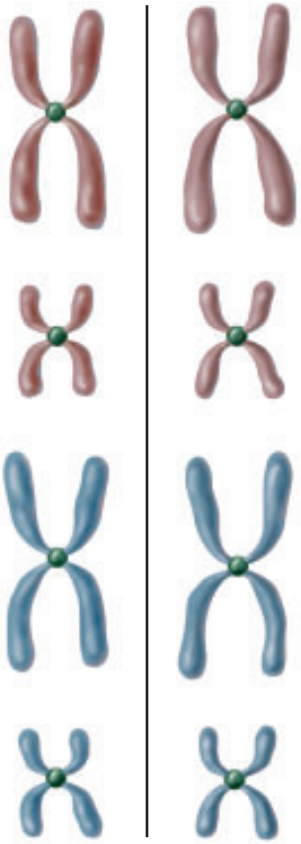

Figure 4

8. Demonstrate crossing over. Break off a piece of clay from one chromosome and attach it to the other chromosome (**Figure 5**). Repeat a few times if you like.
9. To simulate metaphase I, place the chromosomes on either side of the equatorial plate, represented by a line drawn on a piece of paper (**Figure 6**).
10. Choose one of the haploid daughter cells and line the chromosomes up along the equatorial plate. Remove the centromere and move chromosomes to opposite poles (**Figure 7**).

Analysis and Evaluation

Part 1: Mitosis

(a) In step 3, what process did you model?

(b) What do the red and blue pieces of clay represent? What do the green balls of clay represent?

(c) In step 4, what is the diploid chromosome number of the cell?

(d) What phase of mitosis does the model represent?

(e) In step 5, what structure do the single pieces of clay represent after separation?

(f) What phase of mitosis does the model represent?

(g) In step 6, how many chromosomes are in each of the daughter cells?

(h) Compare the daughter cells with the parent cell.

Part 2: Meiosis

(i) In steps 1 to 3, on what basis are chromosomes considered to be homologous?

(j) What is the diploid chromosome number?

(k) In step 8, what must happen before the homologous chromosomes can cross over?

(l) In which phase does crossing over occur?

(m) What happens during crossing over?

(n) In step 9, how does metaphase I of meiosis differ from metaphase of mitosis?

(o) What is the haploid chromosome number?

(p) In step 10, compare the resulting daughter cells of mitosis and meiosis.

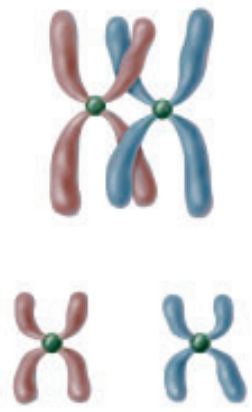

Figure 5

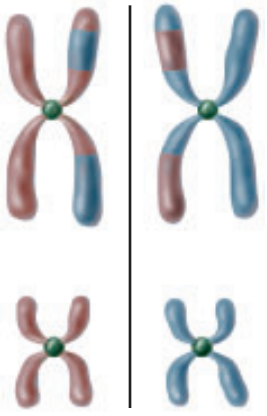

Figure 6

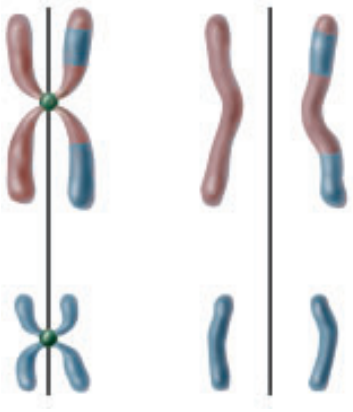

Figure 7

Chapter 17 SUMMARY

Outcomes

Knowledge

- define and explain the significance of chromosome number in somatic and sex cells (i.e., haploidy, diploidy and polyploidy) (17.3, 17.4)
- explain cell cycle events (i.e., interphase, including G1, S, and G2 phases, chromosomal behaviour in mitosis and cytokinesis) (17.1)
- describe spermatogenesis and oogenesis and the reduction of chromosomal number in meiosis (17.3)
- compare the processes of mitosis and meiosis (17.3)
- describe the processes of crossing over and nondisjunction in terms of stages, replication, and resultant chromosome numbers and evaluate their significance to variation in organism inheritance and development (17.4)
- compare the formation of fraternal and identical offspring in a single birthing event (17.1)
- describe the diversity of reproductive strategies by incorporating the principles of mitosis and meiosis when comparing the alternation of generations in a range of organisms (17.3)

STS

- explain that science and technology are developed to meet societal needs and expand human capability (17.2, 17.4)

Skills

- ask questions and plan investigations of questions, ideas, problems, and issues (all)
- gather and record data and information by performing a simulation to demonstrate the behaviour of chromosomes during mitosis (17.1); use a microscope and prepared slides of onion root tip cells to identify the stages of a cell cycle, and calculate the duration of each stage; research and compare a range of reproductive strategies in organisms and present them in charts, tables, or diagrams (17.3)
- analyze data and apply mathematical and conceptual models by preparing and interpreting models of human karyotypes (17.4)
- work as members of a team and apply the skills and conventions of science (all)

Key Terms

17.1

somatic cell
cell cycle
mitosis
cytokinesis
interphase
chromatin
centromere
sister chromatids
centriole
spindle fibre

17.2

enucleated
stem cell
telomere

17.3

meiosis
haploid
diploid
homologous chromosomes
tetrad
synapsis
crossing over
gametogenesis
ootid
polar body
oocyte

17.4

nondisjunction
polyploidy
trisomy
monosomy
karyotype chart

MAKE a summary

1. Sketch the processes of meiosis and mitosis and show the differences between them. Label the sketch with as many of the key terms as possible. Check other sketches and use appropriate designs to make your sketch more clear.
2. Revisit your answers to the Starting Points questions at the start of the chapter. Would you answer the questions differently now? Why?

Go To www.science.nelson.com GO

The following components are available on the Nelson Web site. Follow the links for *Nelson Biology Alberta 20–30*.

- an interactive Self Quiz for Chapter 17
- additional Diploma Exam-style Review Questions
- Illustrated Glossary
- additional IB-related material

There is more information on the Web site wherever you see the Go icon in the chapter.

EXTENSION

CBC radioONE QUIRKS & QUARKS

A Cure for Aging

Dr. Siegfried Hekimi, (professor of biology at McGill University), Dr. Michael West, (Chief Executive Officer of Advanced Cell Technology in Worcester Massachusetts), Dr. Cynthia Kenyon, (biochemistry and biophysics professor from the University of California, San Francisco), and Dr. Marc Tatar (Brown University in Rhode Island) all discuss the causes of aging and their research into slowing the aging process.

www.science.nelson.com GO

Many of these questions are in the style of the Diploma Exam. You will find guidance for writing Diploma Exams in Appendix A5. Science Directing Words used in Diploma Exams are in bold type. Exam study tips and test-taking suggestions are on the Nelson Web site.

www.science.nelson.com GO

DO NOT WRITE IN THIS TEXTBOOK.

Part 1

1. Select the diagram that represents metaphase.

A. Figure 1 (a)
B. Figure 1 (b)
C. Figure 1 (c)
D. Figure 1 (d)

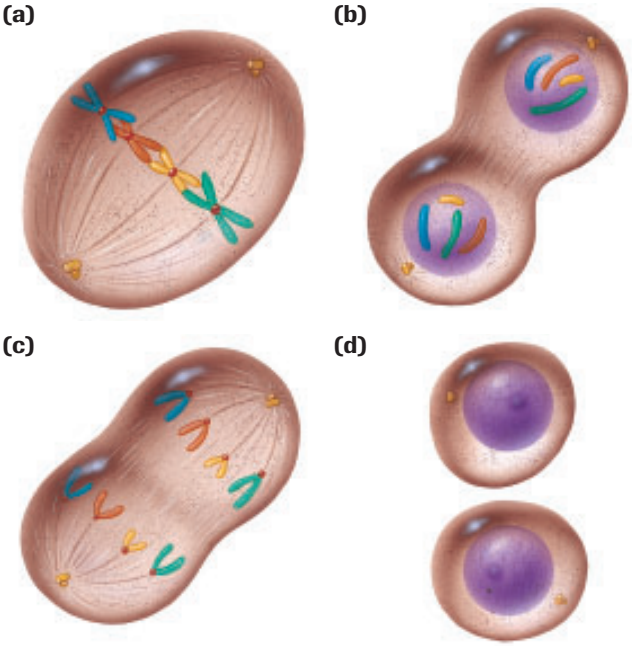

Figure 1

2. NR The following descriptions explain events in the various stages of a cell cycle. Arrange the description in the correct sequence of events. (Record all four digits of your answer.)

1. Chromatids separate and move to opposite poles.
2. Chromosomal alignment occurs in the equatorial plate.
3. Chromosomes become longer and thinner.
4. Chromosomes shorten and thicken.

Use the following information to answer questions 3 to 6.

A student observed three different areas in the mitotic region in an onion root tip. She counted the number of cells that were at each stage of the cell cycle at the time the root was killed and mounted on the slide. Her results are presented in **Table 1**.

Table 1 Number of Cells in Different Stages of Division

Phase	Number of cells			
	Area 1	*Area 2*	*Area 3*	*Total*
interphase	99	79	88	
prophase	12	14	16	
metaphase	6	4	5	
anaphase	0	2	2	4
telophase	2	3	4	9

3. According to the data in **Table 1**, the duration of the phases of the cell cycle, from the longest to the shortest, is

A. prophase, metaphase, anaphase, telophase, interphase
B. interphase, prophase, metaphase, telophase, anaphase
C. interphase, prophase, metaphase, anaphase, telophase
D. not possible to list, since the number of cells and not the duration was observed

4. NR Calculate the percentage of cells in prophase. (Record your answer as a value rounded to one decimal place.)

5. NR If the total time for the completion of one cell cycle is 660 min, determine the time required to complete metaphase. (Record all four digits of your answer.)

6. NR Calculate the percentage of cells undergoing mitosis. (Record your answer as a value rounded to one decimal place.)

7. A researcher studied the growth rate of a malignant cell in mice. Every two days, he counted the number of cells in a 1 mm^2 area, over a period of two months. Select the graph in **Figure 2**, on the next page, that represents the data collected.

A. Figure 2 (a)
B. Figure 2 (b)
C. Figure 2 (c)
D. Figure 2 (d)

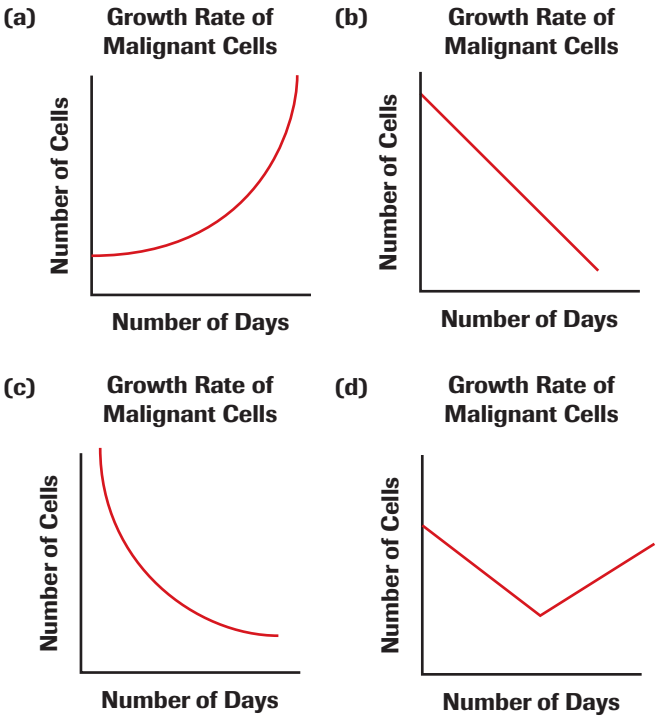

Figure 2

Use the following information to answer questions 8 to 10.

Figure 3 shows the early events in fertilization of a human egg and sperm, and development of the embryo. The numbers refer to the number of chromosomes.

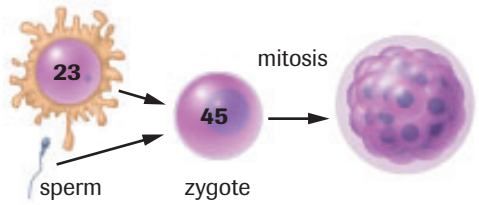

Figure 3

8. Select the number of chromosomes that were in the sperm cell.
A. 20
B. 22
C. 23
D. 45

9. Select the number of homologous pairs of chromosomes that would be in the zygote if it were female.
A. 21
B. 22
C. 23
D. 24

10. Select the number of chromosomes that would be in each blastula cell, following mitosis.
A. 20
B. 22
C. 23
D. 45

11. Indicate which of the following cells would be capable of meiosis:
A. brain cells
B. fat cells
C. cells of a zygote
D. sperm-producing cells of the testes

Part 2

12. **Figure 4** shows plant and animal cells during cell division.
(a) **Identify** each cell as either a plant or an animal cell. **Justify** your answer.
(b) **Identify** the phases of cell division.

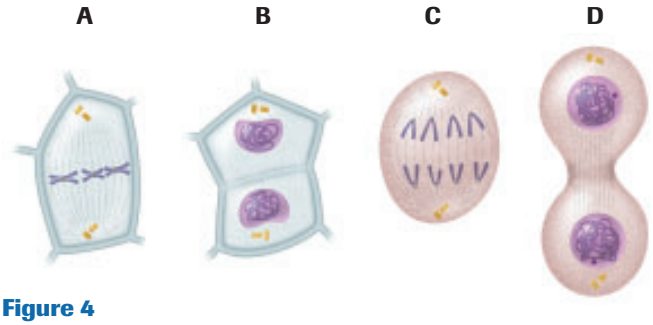

Figure 4

13. **Explain** why a better understanding of the mechanism of cell division may enable scientists to regenerate limbs.

14. **Explain** why the formation of calluses on the hands provides evidence that cell division can be stimulated by cell damage.

15. **Explain** how it is possible to produce a trisomic XXX female.

16. **Sketch** a diagram that shows the kind of nondisjunction that would cause a male and female each with an abnormal number of chromosomes to produce an XYY offspring.

17. If nondisjunction disorders could be eliminated by screening sperm and egg cells, sperm and egg banks could all but eliminate many genetic disorders. **Describe** the social, moral, and ethical implications to society of the systematic elimination of genetic disorders in humans.

Use the following information to answer questions 18 and 19.

Table 2 shows data collected from two different fields of view while examining hamster embryo cells. The number of cells found in each of the cell phases was recorded. It took 660 min to complete one cycle from interphase to interphase.

Table 2

Cell phase	Area 1	Area 2	Total cell count	Time spent in phase
interphase	91	70	?	?
prophase	10	14	?	?
metaphase	2	1	?	?
anaphase	2	1	?	?
telophase	4	4	?	?

18. Copy **Table 2** into your notebook, **determine** the missing values, and complete the table. To calculate the time spent in interphase, for example, you would use the following equation:

$$\frac{\text{Number of cells in interphase}}{\text{Total number of cells counted}} = \frac{\text{Time spent in phase}}{\text{Total time of cycle (660 min)}}$$

19. Using the data provided, **sketch** a circle graph showing the amount of time spent in each phase of the cell cycle.

20. **Identify** one advantage of using a cutting instead of using seeds to grow a new plant.

Use the following information to answer questions 21 to 25.

Fruit flies normally have eight chromosomes. Flies with fewer chromosomes die before maturity. **Figure 5** shows the process of meiosis in three fruit flies.

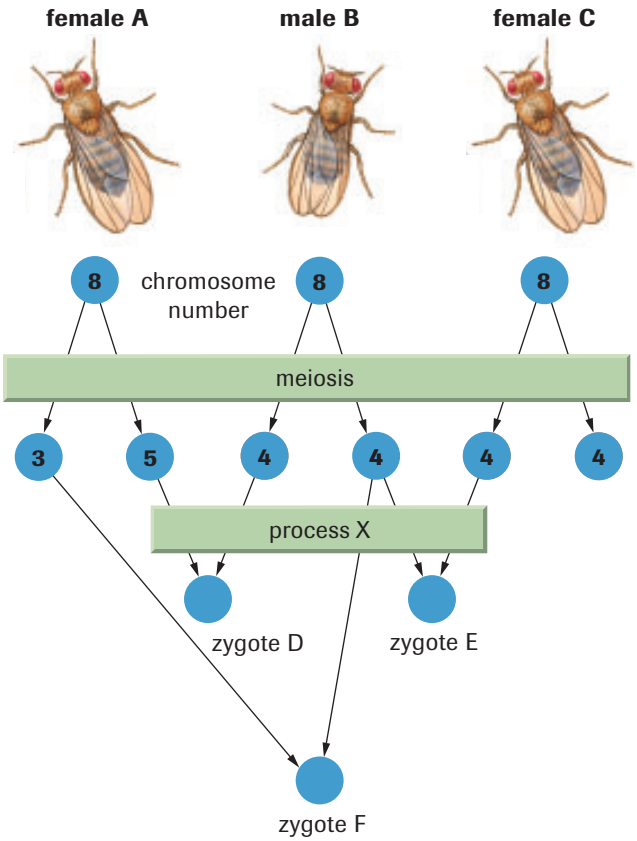

Figure 5

21. **Identify** the parent in which nondisjunction takes place.

22. **Identify** how many chromosomes would be in zygotes D, E, and F.

23. **Describe** what is happening during process X.

24. **Identify** which zygote would most likely be healthy.

25. **Identify** by name the conditions that the other zygotes have.

26. Twins can be either identical or fraternal. Write a unified response that includes the following aspects of twins:

- Copy **Table 3** in your notebook. **Identify** with a check mark (✓) the statements that you believe are always, or almost always, true for fraternal twins and for identical twins.
- **Justify** each choice.

Table 3

Descriptor	Fraternal twins	Identical twins
They have the same blood type.	?	?
They are the same sex.	?	?
They like the same hockey team.	?	?
They have the same mass.	?	?
They have the same hair colour.	?	?
They know what the other one is thinking.	?	?

chapter

18 The Basis of Heredity

In this chapter

- Exploration: Similarities and Differences
- Mini Investigation: Cross-Pollination
- Web Activity: Creating a Personal Profile
- Explore an Issue: Genetic Screening
- Web Activity: Pedigree Analysis
- Investigation 18.1: How Do Environmental Factors Affect Gene Expression?
- Case Study: A Mystery of Blood Types
- Investigation 18.2: Genetics of Corn
- Explore an Issue: Drought-Tolerant and Salt-Tolerant Plants

Have you ever been able to identify a person as a member of a particular family by certain physical traits? Some traits, such as curly hair or a prominent nose, can be traced through a family's lineage. Heredity is the transmission of biological traits from parents to offspring. When the members of different generations all share a particular trait, this is evidence that the trait is inherited. Genetics is the study of inheritance of biological traits.

Biological traits are determined by genes, which are specific segments of DNA. During reproduction, genes of the parent or parents are transmitted to the next generation. Long before we knew of genes and DNA, humans were able to use knowledge of transmission of biological traits to their advantage. Domesticated animals, such as cows and dogs, were produced by choosing parents having traits that were desired in the offspring. Crop plants were also developed by selecting parents with desirable traits.

Every person inherits one of about eight million possible combinations of his or her parents' chromosomes. Your set of genes and your traits are therefore all your own. Even twins who are genetically identical may not share all the same traits.

What patterns can be found in the transmission of genetic traits? How do these relate to the transmission of genes? In this chapter, you will explore patterns of inheritance of biological traits and explain how these patterns arise.

STARTING Points

Answer these questions as best you can with your current knowledge. Then, using the concepts and skills you have learned, you will revise your answers at the end of the chapter.

1. Is it possible for two parents with black hair to have a child with red hair? Why or why not?
2. Sometimes, when breeders cross two individuals with valuable traits, the offspring do not show the same traits. Suggest a reason why this may be so.
3. A team of researchers at the University of Alberta studied sets of identical twins to see if driving a truck or other heavy machinery was related to back pain. Each set of twins included one individual who drove for a living and another who did not. They found that the amount of back pain experienced by a truck-driving twin was the same as for the non-driving twin.
   (a) Why was it important to study identical twins?
   (b) Could the study have used fraternal twins instead? Why or why not?

Career Connections:
Veterinarian; Agrologist

Figure 1
Bobby Hull and Brett Hull starred in the National Hockey League and were the first father and son to win the Hart Trophy.

Figure 2
The father of former prime minister Paul Martin was also a federal politician.

Figure 3
Keifer and Donald Sutherland have successful acting careers.

Exploration — Similarities and Differences

Look at the people shown in **Figure 1**, **Figure 2**, and **Figure 3**. Identify any traits, such as eye colour, eye shape, face shape, and nose length and width, that show a family resemblance. Consider the information in the captions.

- Organize the traits in a chart or table.
- Identify the traits that you think are inherited.

(a) Describe the criteria you used to decide that a trait was inherited.

(b) Brett Hull is one of the NHL's all-time goal scorers. Do you think Brett inherited the ability to score goals from his father, Bobby Hull (**Figure 1**), or is this a skill he learned? Give reasons for your answer.

18.1 Gregor Mendel—Pioneer of Genetics

Figure 1
Gregor Mendel (1822–1884) was an Austrian monk whose experiments with garden peas laid the foundation for the science of genetics.

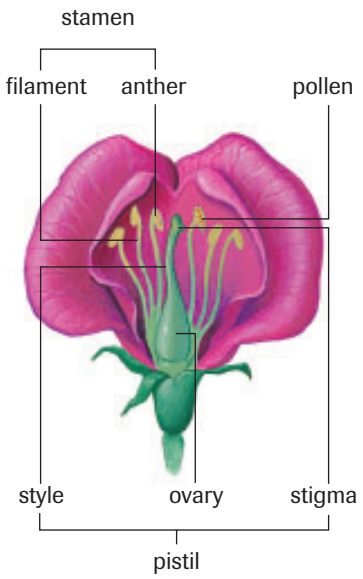

Figure 2
The structure of a flower

progeny new individuals that result from reproduction; offspring

dominant trait a characteristic that is expressed when one or both alleles in an individual are the dominant form

Humans have long understood that certain characteristics were passed down from generation to generation. Stone tablets crafted by the Babylonians 6000 years ago show pedigrees of successive generations of champion horses. However, the first real understanding of inheritance would not come until the work of an Austrian monk, Gregor Mendel, in the mid-19th century (**Figure 1**). Mendel tracked and recorded the transmission of seven visible traits through several generations of the garden pea. To keep track of the different generations, he called the first cross the parental generation, or P generation. The offspring of this cross he called the first filial generation, or the F_1 generation. The next generations were the F_2 generation, the F_3 generation, and so on.

Why did Mendel work with the garden pea? First, he observed that garden peas have a number of characteristics that are expressed in one of only two alternative forms. This made it easier to see which form was inherited.

The second reason is related to how this species reproduces. Garden peas usually reproduce through self-pollination. During pollination, the pollen produced by the anthers of the stamens attaches to the pistil. The pistil consists of the stigma, style, and ovary (**Figure 2**). The ovary contains an egg cell or female sex cell (gamete). Sperm cells (the male gametes) in the pollen grains fertilize the egg cell, and seeds are produced. In self-pollination, the pollen grains and the pistil are from the same plant: in cross-pollination, the pollen grains and the pistil are from different plants. The garden peas that Mendel worked with were "pure" varieties with known traits that came from a long line of self-pollinated pea plants. The traits of each variety had, therefore, been present in all individuals of that variety over many generations.

The Principle of Dominance

When Mendel used pollen from a pea plant with round seeds to fertilize a pea plant with wrinkled seeds, he found that all the offspring (the **progeny**) in the F_1 generation had round seeds. Did this mean that the pollen determines the shape of a seed? Mendel tested this by using pollen from a plant with wrinkled seeds to fertilize a plant with round seeds. Once again, all the progeny had round seeds. Round-seed shape was always the **dominant trait**, regardless of parentage. Mendel called the other wrinkled-seed shape the **recessive trait**. Mendel repeated the experiment for other traits. One trait was always dominant and the other recessive.

Mendel reasoned that each trait must be determined by something he called "factors." Today, we know these factors are genes. Mendel also realized that there can be alternate forms of a gene, which give rise to alternate forms of a trait. We now call the alternate form of a gene an **allele**. For example, the gene for seed colour has two alleles, one that determines green-seed colour and one that determines yellow. Alleles that determine dominant traits are dominant alleles. Alleles that determine recessive traits are recessive alleles. A dominant allele is indicated by an uppercase italic letter, such as *R* for round seeds. The recessive allele is designated by the lowercase italic letter, such as *r* for wrinkled seeds.

Mendel's Principle of Segregation

Mendel next let the F_1 plants self-fertilize, to observe the pattern of transmission of traits in the F_2 generation. When he had crossed pure round-seed plants with pure wrinkled-seed plants, 100 % of the F_1 generation had round seeds. Mendel was astonished to find that 75 % of the F_2 generation had round seeds and 25 % had wrinkled seeds. That is, for seed shape, the ratio was 3:1 round to wrinkled. He performed crosses to follow other traits and found the F_1 generations all had the same 3:1 ratio of dominant to recessive trait.

recessive trait a characteristic that is expressed only when both alleles in an individual are the recessive form

allele one of alternative forms of a gene

To explain these ratios, Mendel reasoned that each plant must carry two copies (alleles) of each gene that can be the same or different. An individual with round seeds must carry at least one dominant allele (*R*), but individuals with wrinkled seeds must always carry two copies of the recessive allele (*rr*).

When both alleles of a gene pair are the same, an individual is said to be **homozygous** for that trait. When the alleles of a gene pair are different, an individual is **heterozygous** for that trait. The complement of genes of an organism is called its **genotype**, and the physical expression of the genotype is the **phenotype**.

homozygous having identical alleles for the same gene

heterozygous having different alleles for the same gene

genotype the genetic complement of an organism

phenotype the observable characteristics of an organism

Mendel also correctly concluded that the two copies of a gene in a gene pair undergo **segregation** during the formation of the sex cells. Each mature gamete contains only one member of a gene pair. When an individual is homozygous for a gene, all of its gametes carry the same allele. When an individual is heterozygous for a gene, each gamete could receive either allele. **Figure 3 (a)** shows the results of a cross between two homozygous peas. At fertilization, the new individual receives one copy of the gene from the female parent and one from the male parent. All members of the F_1 generation, therefore, are heterozygous. When the F_1 generation was allowed to self-pollinate, three different genotypes were produced, which determined the two phenotypes that Mendel observed (**Figure 3 (b)**).

segregation the separation of alleles during meiosis

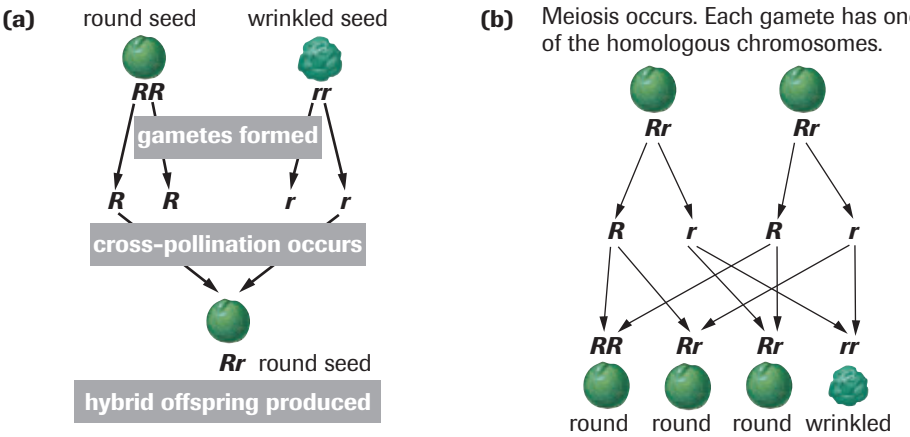

Figure 3

(a) When a pea plant homozygous for round seeds is cross-pollinated with a pea plant homozygous for wrinkled seeds, the offspring are all heterozygous.

(b) The F_2 progeny from a cross of two heterozygous pea plants with round seeds will have three possible genotypes *RR*, *Rr*, and *rr*.

EXTENSION

Genetic Terms

This animation gives a visual review of some of the terms used in studying genetics.

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mini Investigation Cross-Pollination

Materials: two plants of the same species that have different colours of flowers, small scissors, paint brush, plastic bags, potting soil, water, small pots

- On the plant you want to be the seed-parent, select a flower that is not yet open. Using **Figure 4** as a guide, remove the anthers from the flower.
- Using the paint brush, transfer pollen from the pollen-parent to the stigma of the seed-parent flower from which you removed the anthers.

(a) Predict the flower colour of the offspring of your cross-pollinated plant. Give reasons for your prediction.

(b) Why were the anthers removed from the plant that received the pollen?

(c) Why was a plastic bag placed over the flower?

- If there is time, collect and grow seeds from the flower you pollinated. Cover the pollinated flower with a plastic bag. Once the flower has produced seeds, plant the seeds in moist soil. Place the plant in sunlight (or under a bright light) and keep it watered until it produces flowers.

(d) Was your prediction correct?

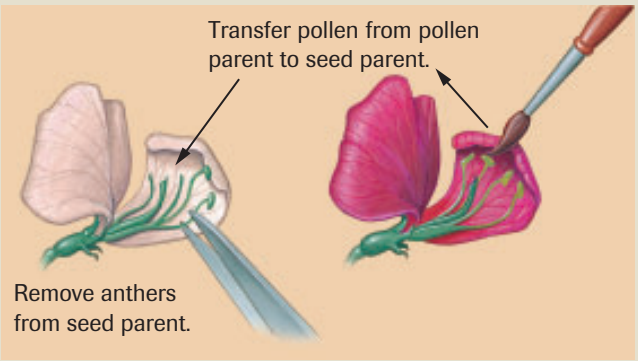

Figure 4
Pollen is transferred from the donor plant to the pistil of the recipient, which has had its stamens removed to prevent self-pollination.

SUMMARY Gregor Mendel—Pioneer of Genetics

- Inherited traits are controlled by factors—genes—that occur in pairs. Each member of a pair of genes is called an allele.
- One factor, or allele, masks the expression of another. This is known as the principle of dominance.
- A pair of factors, or alleles, separates from one another (segregate) during the formation of sex cells. This is often referred to as the law of segregation.

Section 18.1 Questions

1. Why were the pea plants selected by Mendel ideally suited for studying the transmission of traits?
2. Explain why, under normal circumstances, an individual can carry only two alleles of a gene.
3. Use an example that helps differentiate between the terms genotype and phenotype.
4. Black fur colour is dominant to yellow in Labrador retrievers.
   (a) Explain how the genotype of a homozygous black dog differs from that of a heterozygous black dog.
   (b) Could the heterozygous black dog have the same genotype as a yellow-haired dog? Explain.
5. A pea plant with round seeds is cross-pollinated with a pea plant that has wrinkled seeds. The plant with round seeds is heterozygous. Indicate each of the following:
   (a) the genotypes of the parents
   (b) the gametes produced by the parent with round seeds
   (c) the gametes produced by the parent with wrinkled seeds
   (d) the possible genotype(s) and the phenotype(s) of the F_1 generation

Probability and Inheritance of Single Traits 18.2

For every cross, Mendel kept track of the number of offspring that inherited the dominant trait and recessive trait. Based on mathematical analysis of these numbers, Mendel also concluded that each gamete produced by a heterozygous individual has an equal chance of getting either allele of a gene pair. Recall that when Mendel allowed peas that were heterozygous for the seed shape allele to self-pollinate, 75 % of the F_2 generation had the round-seed phenotype and 25 % had the wrinkled-seed phenotype. In other words, the **phenotypic ratio** of offspring with the dominant trait to offspring with the recessive trait was 3 to 1. To get this ratio, each sex cell must have had an equal probability of getting the *R* allele as the *r* allele during the process of segregation.

The probability of an outcome is a measure of the likelihood that the outcome will occur. Probability may be expressed as a fraction, a decimal, or a percentage. Probability (*P*) can be determined using the following formula:

$$P = \frac{\text{number of ways that a given outcome can occur}}{\text{total number of possible outcomes}}$$

For example, you might calculate the probability of getting heads when you toss a coin. There is only one way of tossing heads, so the numerator is 1. Since there are two possible outcomes in total, the denominator is 2. Therefore, the probability *P* of tossing heads is $\frac{1}{2}$, or 0.5, or 50 %.

A **Punnett square** is a chart that can help us to predict the phenotypes of the progeny of a cross between parents of known genotypes, or to deduce the genotypes of parents from the observed phenotypic ratio of their progeny. Punnett squares also allow us to determine the expected ratio of the genotypes (**genotypic ratio**) and the phenotypes for a cross, and to state the probability of that particular genotype or phenotype will occur in the progeny of a cross.

phenotypic ratio the ratio of offspring with a dominant trait to the alternative, recessive trait

Punnett square a chart used to determine the predicted outcome of a genetic cross

genotypic ratio the ratio of offspring with each possible allele combination from a particular cross

SAMPLE exercise 1

A breeder crosses a pea plant with wrinkled seeds and a pea plant with round seeds. She knows that the plant with round seeds is heterozygous for the gene for seed shape. The allele for round seeds (*R*) is dominant over the allele for wrinkled seeds (*r*). Determine the expected genotypic ratio and phenotypic ratio of the progeny.

Solution

Since *r* is the recessive allele, the genotype of the plant with wrinkled seeds must be *rr*. Since the plant with round seeds is heterozygous, its genotype must be *Rr*. The symbols for the alleles in the gametes are written across the top and along the left side of the Punnett square (**Figure 1**). Each cell is then filled in by entering one allele from the top of the square and a second allele from the side of the square.

Figure 2 shows a completed Punnett square for a cross between a heterozygous round-seed pea plant and a wrinkled-seed pea plant. Two of the four cells show the genotype *Rr* and two show *rr*. The expected genotypic ratio in the progeny of *Rr* to *rr* is, therefore, 1:1. Offspring with genotype *Rr* will have round seeds, and those with genotype *rr* will have wrinkled seeds.

Therefore, the phenotypic ratio is 1:1 ($\frac{1}{2}$ round and $\frac{1}{2}$ wrinkled).

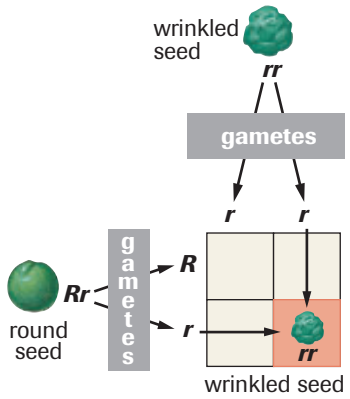

Figure 1
The partially completed Punnett square for a cross between a pea plant with genotype *rr* and a pea plant with genotype *Rr*. The genotype *rr* in one cell of the Punnett square is one of four possible combinations of the parental alleles.

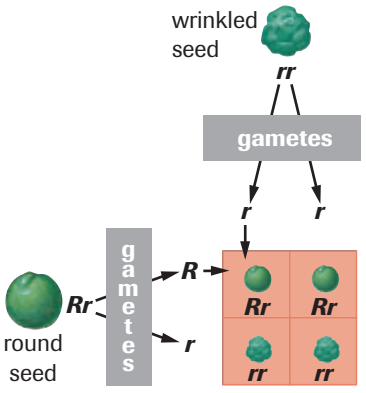

Figure 2

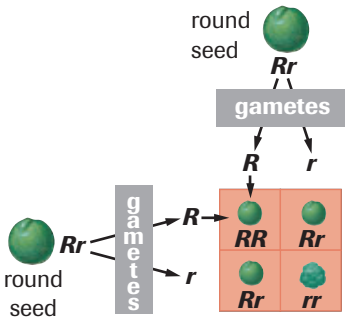

Figure 3
A Punnett square showing the results of a cross between two heterozygous plants with round seeds

SAMPLE exercise 2

For the cross shown in **Figure 3**, what is the probability that an offspring will have a phenotype of wrinkled seeds? Express the answer as a percent.

Solution

Since the allele for wrinkled seeds, *r*, is recessive, only offspring with a genotype *rr* will have wrinkled seeds. From the Punnett square, 1 of every 4 offspring are expected to have this genotype, so the probability that an offspring will have wrinkled seeds is 25 %.

Practice

1. What is the phenotypic ratio of the cross in the Punnett square shown in **Figure 4**?

	T	T
t	Tt	Tt
t	Tt	Tt

Figure 4
Punnett square of a monohybrid cross between a homozygous tall pea and a homozygous short pea

2. Using a Punnett square, determine the expected phenotypic ratio and genotypic ratio for the progeny of a cross between a pea plant that is homozygous for the white allele (*r*) for flower colour and a pea plant that is homozygous for the red allele (*R*).

EXTENSION

F_2 Ratios
This animation shows some of the results of Mendel's crosses, which you can then convert to phenotypic ratios. How close are the observed phenotypic ratios to the predicted phenotypic ratio?

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WEB Activity

Case Study—Creating a Personal Profile

Some human genes determine visible traits that show an inheritance pattern that is similar to that of Mendel's garden peas. As a result, you can predict a person's genotype for these traits just by observing him or her. In this activity, you will use a list of some common dominant and recessive traits, and use this information to create a profile of your own phenotype and potential genotype.

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EXTENSION

Genetics
In this Virtual Biology Laboratory, you can assess data and perform simulated crosses to explain the inheritance of shell colour in glyptodonts, an extinct relative of the armadillo.

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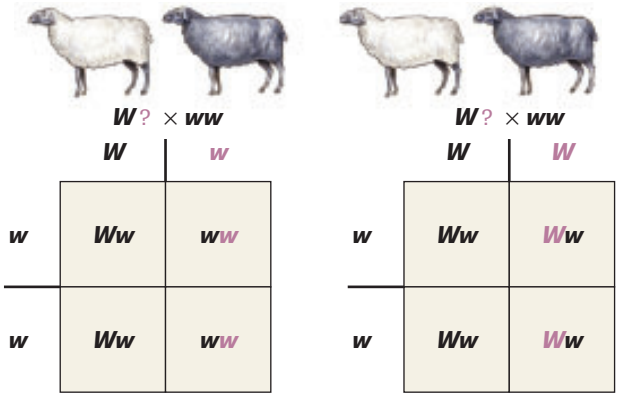
Test Crosses

Wool producers often prefer sheep with white wool, since black wool tends to be brittle and difficult to dye. Black sheep can be avoided by breeding only homozygous white rams. However, the allele for white wool *(W)* is dominant over the allele for black wool *(w)*, so white rams can be heterozygous. How could a wool producer be sure that a white ram is homozygous?

A **test cross** is the cross of an individual of unknown genotype to an individual with a recessive genotype. The phenotypes of the F_1 generation of a test cross reveal whether an individual with a dominant trait (such as a white ram) is homozygous or heterozygous for the dominant allele. If a white ram is crossed with a black ewe and the observed phenotypic ratio is 1:1 black to white, then the genotype of the ram must be *Ww* (**Figure 5**). If all the offspring are white, then the genotype of the white ram must be *WW*.

Test crosses are the simplest way of determining the genotype of an individual. Sometimes, however, the parents are not available to test cross. When only information about the phenotypes of the offspring of a cross is available, the genotypes and phenotypes of the parents can be found by working backwards through a Punnett square.

test cross the cross of an individual of unknown genotype to an individual that is fully recessive



Half of the offspring are black and half are white.

All of the offspring are white.

Figure 5
A test cross is a way of determining if an individual with the dominant trait is heterozygous or homozygous.

SAMPLE exercise 3

A horticulture worker has seeds from a particular cross, but has no information about the genotype or the phenotype of the parents. He plants and grows the offspring, and records the traits of each offspring **(Table 1)**. What was the genotype and phenotype of the parent plants?

Table 1

Offspring phenotype	Numbers
round-seed peas	5472
wrinkled-seed peas	1850

Solution

Determine the observed phenotypic ratio of the progeny, rounding off if needed.

$$\frac{\text{round}}{\text{wrinkled}} = \frac{5472}{1850} \approx \frac{3}{1}$$

List the possible genotypes for each phenotype, as shown in **Table 2**.

Table 2

Phenotype	Genotype
round-seed peas	*RR* or *Rr*
wrinkled-seed peas	*rr*

EXTENSION

Factors that Contribute to Genetic Variation

In this Audio Clip, you will hear about the underlying mechanisms that create genetic variation in the offspring of sexually reproducing individuals.

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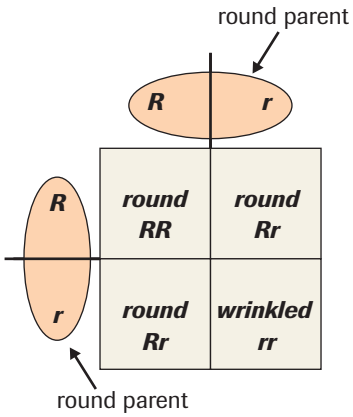

Figure 6
The observed phenotypic ratio is the same as the ratio predicted by the Punnett square.

A 3:1 phenotypic ratio occurs when two heterozygous individuals are crossed, so we know that the parents must be heterozygous. Since only $\frac{1}{4}$ of the progeny had wrinkled seeds, this is the recessive phenotype and must be determined by two copies of the recessive allele. The parents were heterozygous, so their genotype was *Rr*. Check the answer using a Punnett square **(Figure 6)**.

Practice

3. A fish breeder has a red male cichlid of unknown parentage. Red colour is dominant to yellow in the fish. He must know whether the fish is heterozygous for these colours. Suggest a way the fish breeder might find out the genotype of his red male. Use a Punnett square to explain your answer.

4. A neighbour gives a home gardener some seeds that he collected last year from his red carnations. The gardener plants 50 of the seeds and is surprised to find 12 of the plants have white flowers. Assuming that all the seeds came from one cross, what was the genotype of the parents?

SUMMARY Probability and Inheritance of Single Traits

- By using a Punnett square, the expected phenotypic ratio and genotypic ratio of the offspring of a cross can be determined.
- Probability, $P = \dfrac{\text{number of ways that a given outcome can occur}}{\text{total number of possible outcomes}}$

  Probability values can be used to predict the likelihood that a particular phenotype will appear in a cross.
- A test cross is the cross of an individual of unknown genotype to an individual with a fully recessive genotype.

Section 18.2 Questions

1. In Dalmatian dogs, the spotted condition is dominant to non-spotted.
   (a) Using a Punnett square, show the cross between two heterozygous parents.
   (b) A spotted female Dalmatian dog has six puppies sired by an unknown male. From their appearance, the owner concludes that the male was a Dalmatian. Three of the pups are spotted and three are not. What is the genotype and phenotype of the puppies' father?

2. For Mexican hairless dogs, the hairless trait is dominant to hairy. A litter of eight pups is found; six are hairless and two are hairy. What are the genotypes of their parents?

3. Test crosses are valuable tools for plant and animal breeders.
   (a) Provide two practical examples of why a cattle rancher might use a test cross.
   (b) Why are most test crosses performed using bulls rather than cows?

Pedigree Charts 18.3

Pedigree analysis is another tool for solving genetic problems. This approach is especially useful when it is not possible to perform crosses using specific individuals or to generate large numbers of progeny, such as for humans. A **pedigree chart** is like a family tree in which the inheritance of a trait can be traced from parents to offspring.

pedigree chart a chart used to record the transmission of a particular trait or traits over several generations

A pedigree chart shows the family relationship among individuals. Symbols identify the gender of each individual and whether an individual had the trait of interest. Pedigree charts may also show when an individual is known to be homozygous or heterozygous for a trait. The top of **Figure 1** shows some commonly used symbols. The pedigree chart in the lower part of **Figure 1** shows the transmission of an inherited disease among members of a family. Genetic counsellors may use pedigree charts in their work.

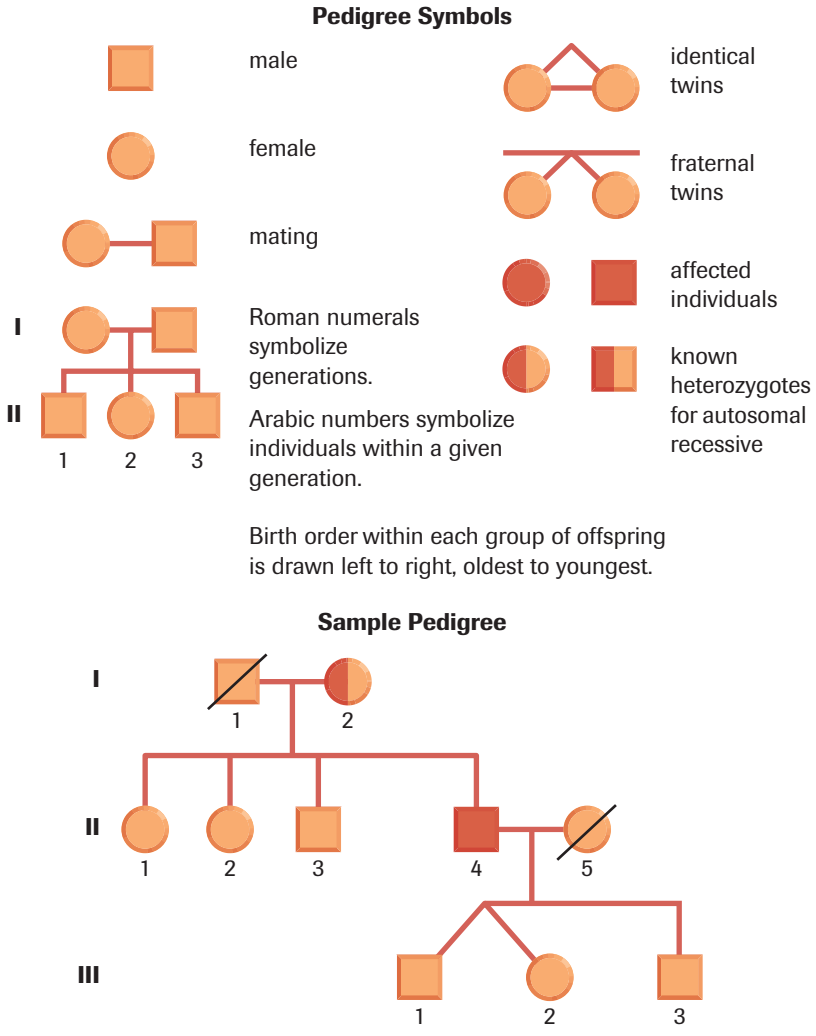

Figure 1

Squares represent males and circles represent females. A slash through a symbol indicates that person is deceased. Vertical lines connect parents to offspring, horizontal lines connect mates and connect siblings. Individuals affected by the inherited disease are identified by the darker-coloured symbols. Symbols having two different colours identify individuals heterozygous for the disease.

Practice

1. People with albinism do not produce normal pigment levels. Albinism is a recessive trait. Use the pedigree chart in **Figure 2** to answer the following questions. Use an uppercase "A" to represent the dominant allele, and a lowercase "a" for the recessive allele.
   (a) How many children do the parents A and B have?
   (b) Indicate the genotypes of the parents.
   (c) Give the genotypes of M and N.

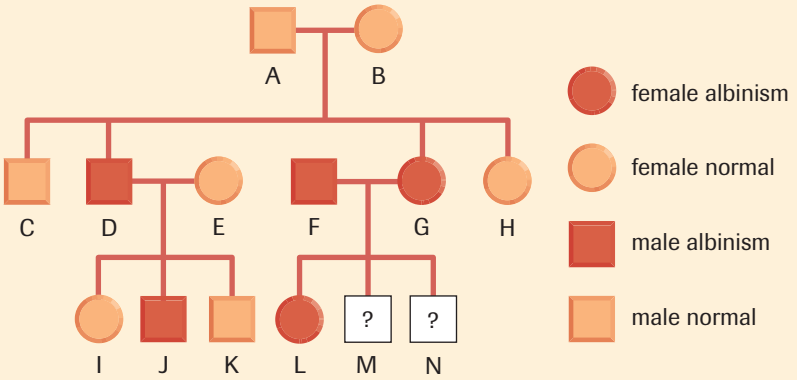

Figure 2

EXPLORE an issue

Genetic Screening

Due to advances in technology, it is now possible to get information about the genotype of any person relatively easily. Genetic screening may be carried out before birth (prenatal screening) or any time after birth. The most common reason for parents to want prenatal genetic screening is because they are at increased risk of passing a genetic disease to their child.

Thalassemia is one genetic disease for which prenatal genetic screening may be performed. Thalassemia is a disease of the blood, which affects a person's ability to produce enough red blood cells. Only people with two copies of a mutant allele of a particular a gene will have the disease. Genetic screening for thalassemia is performed only on those people with a family history of the disease. Prenatal screening can identify the presence of the thalassemia allele before the child is born.

Persons at risk of Huntington disease may request either pre- or post-natal screening. Huntington disease is a neurological disorder caused by a dominant allele. Huntington is characterized by rapid deterioration of nerve control, which causes a range of symptoms, including involuntary movements, slurred speech, loss of memory, and depression. Huntington disease is fatal. There is no cure and available treatments have little effect on symptoms. Symptoms of Huntington disease begin gradually, usually in middle age, when most people have already had children. Genetic screening allows people to know whether they have inherited the disease before any symptoms develop, so they may know whether they are at risk of passing it on to their children.

Issue Checklist

● Issue	○ Design	● Analysis
● Resolution	● Evidence	● Evaluation

Understanding the Issue

- Working in a group, conduct research and find out more about genetic screening.

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1. Define genetic screening. Describe some technologies used in genetic screening.
2. What are some advantages of genetic screening? Provide an example.
3. What are some physical dangers associated with genetic screening methods? Provide an example.

Take a Stand

Consider this position statement: Genetic screening should be compulsory for any person with a family history of a genetic disease.

With your group members, create a list of different stakeholders in this issue. Based on your research, determine points that support and refute the position statement from the perspective of each stakeholder. Then, decide whether your group agrees or disagrees with the position statement. Present your position to the class. Prepare to defend your group's position in a class discussion.

WEB Activity

Simulation—Pedigree Analysis

Complete the interactive Pedigree Analysis Tutorial in this Virtual Biology Laboratory. You can also use pedigree analysis to examine the inheritance of several genetic diseases in humans, and to act as a "genetic counsellor" in some hypothetical case studies.

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SUMMARY Pedigree Charts

- A pedigree chart traces the inheritance of a trait from parents to offspring through several generations.
- Pedigree charts are useful in cases when it is not possible to perform and follow specific crosses, such as in human genetic studies.

Section 18.3 Questions

1. A woman begins to show symptoms of Huntington disease. Her father had Huntington disease, but her mother never developed the disorder. Neither her husband nor anyone in his immediate family have any symptoms.
(a) What is the genotype of the woman with Huntington disease?
(b) What is the probable genotype of the woman's husband?
(c) If the woman has six children, how many are likely to develop Huntington disease?

2. Phenylketonuria (PKU) is a genetic disorder caused by a dominant allele. Individuals with PKU are unable to metabolize a naturally occurring amino acid, phenylalanine. If phenylalanine accumulates, it inhibits the development of the nervous system, leading to mental retardation. The symptoms of PKU are not usually evident at birth, but can develop quickly if the child is not placed on a special diet. The pedigree in **Figure 3** shows the inheritance of the defective PKU allele in a family.
(a) How many generations are shown by the pedigree?
(b) How many children were born to the parents of the first generation?

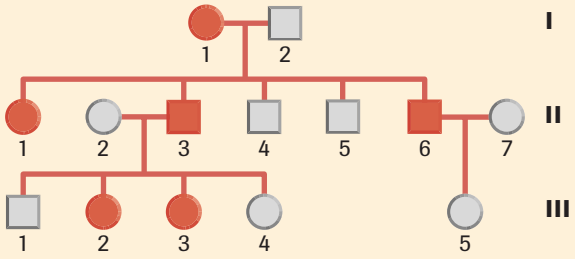

Figure 3

(c) What is the genotype of individuals 1 and 2, generation I?
(d) How is it possible that in generation II, some of the children showed symptoms of PKU, while others did not? (Hint: Use a Punnett square to help with your explanation.)
(e) For individuals 6 and 7, in generation II, a child without PKU symptoms was born. Does this mean that they can never have a child with PKU? Explain your answer.

3. Research the inheritance of one of the traits in **Table 1** in a family that you know. Get information from at least three generations of the family. Use the information you collect to make a pedigree chart.

Table 1

Trait	Dominant	Recessive
freckles	present	absent
dimples	present	absent
earlobe	suspended	attached
hairline	pointed on forehead	straight across forehead
chin dimple	present	absent

4. (a) How or where might genetic screening be used for purposes other than genetic counselling?
(b) What laws, if any, do you think are likely to arise regarding the use of genetic screening? Why?

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18.4 Other Patterns of Inheritance

pleiotropic gene a gene that affects more than one characteristic

EXTENSION

Pleiotropic Effects of Marfan Syndrome

Marfan Syndrome is caused by a mutation in a single gene. This animation shows you how this one gene affects four different organ systems.

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wild type the most common allele of a gene with multiple alleles

mutant any allele of a gene other than the wild type allele

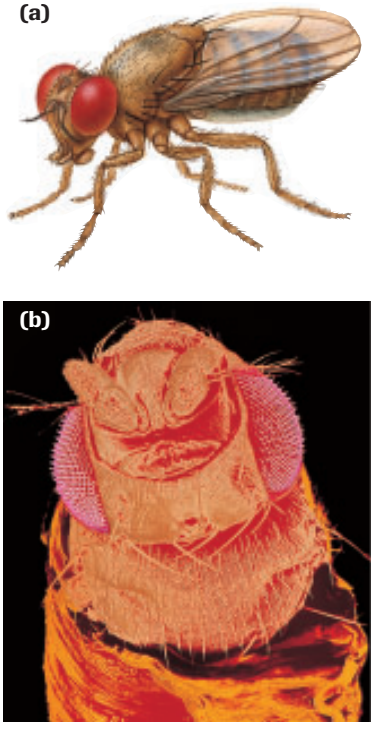

Figure 1

(a) *Drosophila melanogaster*, the fruit fly, is widely used for genetic studies.

(b) Wild type, or red, is the most common eye colour. It is dominant over all the other alleles for eye colour.

The traits that Mendel studied showed little variability. Each had only two alleles, one that was clearly dominant and one clearly recessive. However, many inherited traits show more variability than just two alternate forms. These types of traits will not be inherited in the predicted 3:1 phenotypic ratio of a trait with one dominant allele and one recessive allele.

Pleiotropic Genes

Some genes, called **pleiotropic genes**, affect many different characteristics. Sickle-cell anemia, a blood disorder, is caused by a pleiotropic gene. Normal hemoglobin (the pigment that carries oxygen in the blood) is produced by the allele *HbA*. Sickle cell anemia occurs in individuals who have two copies of the mutated allele, *HbS*. This mutation produces abnormally shaped hemoglobin molecules that interlock with one another. The new arrangement of molecules changes the shape of the red blood cells, which become bent into a sickle shape. The sickle-shaped red blood cells cannot pass through the capillaries, and so cannot deliver oxygen to the cells. People with sickle-cell anemia can suffer from fatigue and weakness, an enlarged spleen, rheumatism, and pneumonia. Patients often show signs of heart, kidney, lung, and muscle damage.

Multiple Alleles

When traits are determined by more than two (multiple) alleles, the most commonly seen trait is called the **wild type**, and the allele that determines it is the wild-type allele. Non-wild-type traits are said to be **mutant**, and the alleles that determine them are mutant alleles. In most cases of multiple alleles, there is a hierarchy of dominance.

Members of the species *Drosophila melanogaster*, the fruit fly (**Figure 1**), can have any one of four eye colours. Red eye colour is the wild type, but the eyes may also be apricot, honey, or white. The *Drosophila* species as a whole has more than two alleles for eye colour but, since fruit flies are diploid, each individual carries only two genes for eye colour.

The dominance hierarchy and symbols for eye colour in *Drosophila* are shown in **Table 1**. When there are multiple alleles for the same gene, upper case letters and superscript numbers are used to express the dominance relationships between the different alleles. For simplicity, the capital letter *E* is used for the eye colour gene and superscript numbers to indicate the position of each allele in the dominance hierarchy.

Table 1 Dominance Hierarchy and Symbols for Eye Colour in *Drosophila*

Phenotype	Allele symbol	Possible genotype(s)	Dominant over
wild type	E^1	E^1E^1, E^1E^2, E^1E^3, E^1E^4	apricot, honey, white
apricot	E^2	E^2E^2, E^2E^3, E^2E^4	honey, white
honey	E^3	E^3E^3, E^3E^4	white
white	E^4	E^4E^4	

SAMPLE exercise 1

What will be the phenotypic ratio of the offspring from the mating of the following *Drosophila* individuals?

E^1E^4 (wild-type eye colour) × E^2E^3 (apricot eye colour)

Solution

The problem can be solved by using a Punnett square. The first parent is heterozygous, and so will produce gametes with the E^1 allele and the E^4 allele. The other parent is also heterozygous, and will produce gametes with the E^2 allele and the E^3 allele. The Punnett square for this cross is, therefore, as shown in **Figure 2**.

Using the dominance hierarchy in **Table 1**, the phenotypic ratio of the F_1 offspring will produce two wild-type eye colour (genotypes E^1E^2 and E^1E^3) to one apricot eye colour (genotype E^2E^4) to one honey eye colour (genotype E^3E^4).

Practice

1. A student working with *Drosophila* makes the following cross:

   E^1E^2 (wild-type eye colour) × E^2E^4 (apricot eye colour)

   What will be the phenotypic ratio of the offspring?

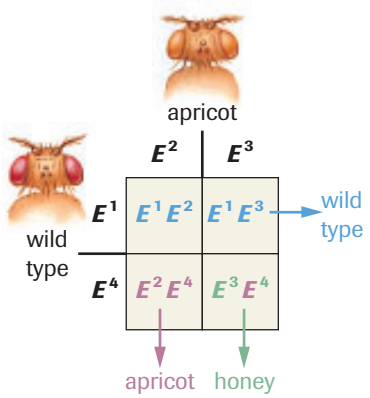

Figure 2
A cross between a fruit fly with wild-type eye colour and one with apricot-coloured eyes

Incomplete Dominance

When two alleles are equally dominant, they interact to produce a new phenotype—this form of interaction between alleles is known as **incomplete dominance**. When an individual is heterozygous for two alleles that show incomplete dominance, both alleles are equally expressed, but at half the level that would occur were the individual homozygous for either allele. The phenotype of a heterozygous individual is, therefore, intermediate between its homozygous parents. For example, when a homozygous red snapdragon is crossed with a homozygous white snapdragon, all of the F_1 generation have pink flowers. If members of the F_1 generation are crossed, the F_2 generation has a surprising phenotypic ratio of one red to two pink to one white (1:2:1). The Punnett square in **Figure 3** shows the genotypes behind this ratio.

incomplete dominance the expression of both forms of an allele in a heterozygous individual in the cells of an organism, producing an intermediate phenotype

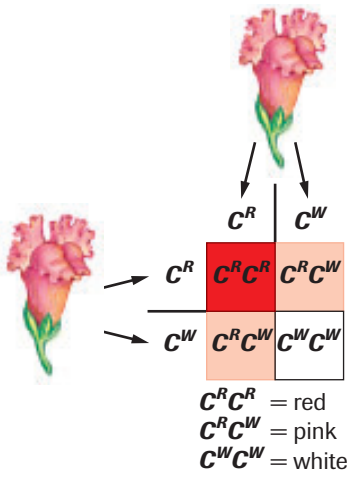

Figure 3
Colour in snapdragons is an example of incomplete dominance. When homozygous red-flowered snapdragons are crossed with homozygous white-flowered snapdragons, the F_1 generation all have pink flowers. When a cross is made between two F_1 individuals, the F_2 generation has a phenotypic ratio of one red to two pink to one white.

Codominance

codominance the expression of both forms of an allele in a heterozygous individual in different cells of the same organism

Another form of allele interaction is **codominance**. When two alleles show codominance, both alleles are fully expressed in a heterozygous individual, but not in the same cells. Coat colour in shorthorn cattle shows codominance (**Figure 4**). Red coats are composed of all red hairs, and white coats are all white hairs. When a red shorthorn is crossed with a white shorthorn, any calves produced will have roan-coloured coats, which is intermediate between the red and the white coat colour. However, each hair is not the intermediate roan colour. Instead, a roan coat has a mixture of white hairs and red hairs.

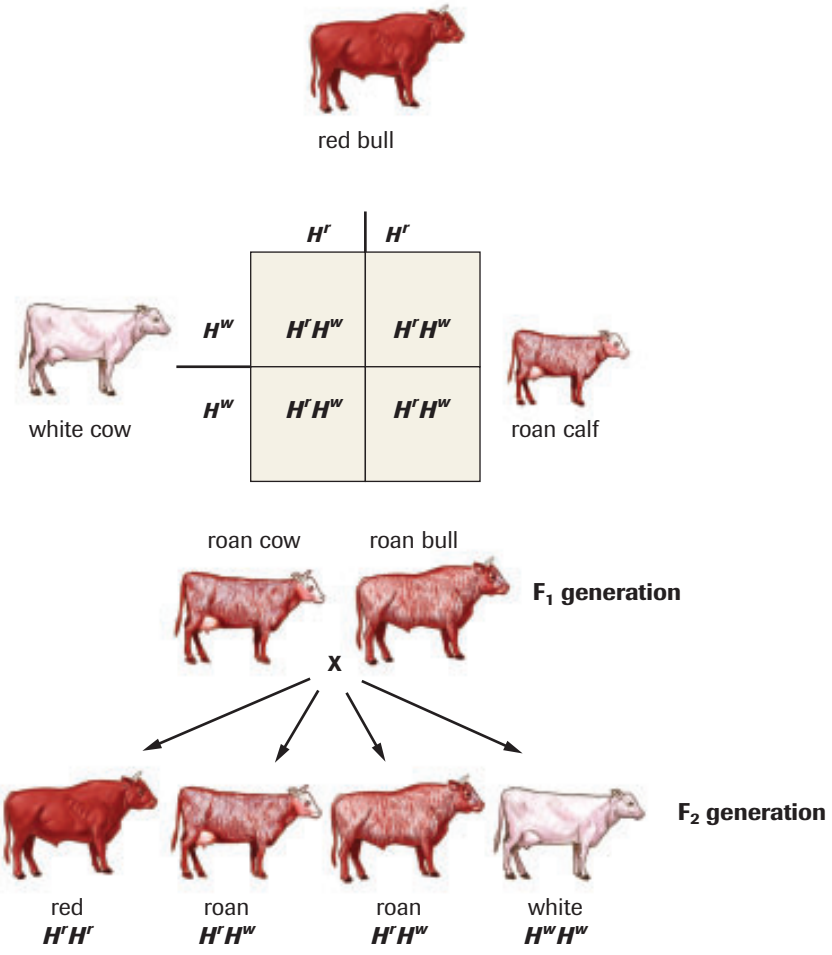

Figure 4
In codominance, either one of two different alleles is expressed. In shorthorn cattle, the coats of roan animals have intermingled red and white hair.

CAREER CONNECTION

Veterinarian

Veterinarians provide health care services that include the diagnosis and treatment of injured and sick animals. They give advice about the breeding of animals and perform genetic procedures and embryo transfers. Veterinarians work long hours and are dedicated animal health specialists. Learn more about their duties.

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Environment and Phenotype

Sometimes, variation of a trait is determined by the interaction of the genotype with the environment. The environment can have a profound effect on phenotype. Himalayan rabbits have black fur when they are raised at low temperatures, but white fur when raised at high temperatures. In some cases, different parts of the same organism can have different traits when exposed to different environments. Leaves of the water buttercup, *Ranunculus aquatilis*, that develop above the surface of the water are broad, lobed, and flat, while those that develop below the water are thin and finely divided. However, the leaves all have identical genetic information.

EXTENSION

Coat Colour in the Himalayan Rabbit

View this animation of how coat colour in this species is affected by temperature.

www.science.nelson.com GO

INVESTIGATION 18.1 *Introduction*

How Do Environmental Factors Affect Gene Expression?

Design and carry out an investigation of the effect of an environmental factor on the phenotype of genetically identical plants.

Report Checklist

- Purpose
- Problem
- Hypothesis
- Prediction
- Design
- Materials
- Procedure
- Evidence
- Analysis
- Evaluation
- Synthesis

To perform this investigation, turn to page 620.

Case Study

A Mystery of Blood Types

Humans have four blood types; A, B, AB, and O. The alleles for blood types A and B are codominant but dominant to O (**Table 2**). We also each have one of two forms of rhesus factor—the positive form (Rh+) or the negative form (Rh−). The allele for the Rh+ form is dominant to the Rh− allele. Blood types can identify individuals and family members.

Table 2 Human Blood Types

Phenotypes	Genotypes
Type A	I^AI^A, I^Ai
Type B	I^BI^B, I^Bi
Type AB	I^AI^B
Type O	ii

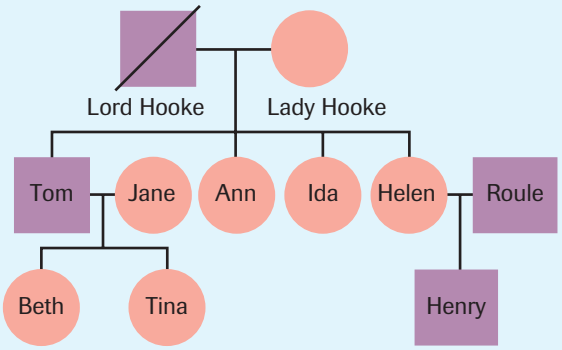


Figure 6
The family tree of the members of Lord Hooke's family who were in the castle

Evidence

In Black Mourning Castle, a scream echoed from the den of Lord Hooke. When the maid peered through the door, a freckled arm reached for her neck. She bolted and telephoned the police. Inspector Holmes arrived to find the dead body of Lord Hooke. Apparently, the lord had been strangled. The inspector noted blood on a letter opener, even though Lord Hooke did not have any cuts. This blood was type O, Rh−. Inspector Holmes took blood samples from the family members shown in **Figure 6**.

The inspector gathered the information shown in **Table 3**. The gene for freckles is dominant to the gene for no freckles. Some family members were wearing long-sleeved shirts, so the inspector could not determine whether freckles were present.

The inspector then announced, "Lady Hooke had been unfaithful to her husband. One of the heirs to the fortune was not Lord Hooke's child. The murder was committed to preserve a share of the fortune!"

Table 3 Traits of the Hooke Family

Family	Blood type	Rh factor	Freckles
Lord Hooke	AB	+	no
Lady Hooke	A	+	no
Helen	A	+	no
Roule	O	+	no
Henry	refused blood test		?
Ida	A	–	?
Ann	B	+	?
Tom	O	–	no
Jane	A	+	?
Beth	O	–	?
Tina	A	+	yes

Case Study Questions

1. Who was the murderer? What was the murderer's probable blood type?
2. Describe how you obtained your answer.
3. How did the inspector eliminate the other family members?

SUMMARY Other Patterns of Inheritance

- Some genes have more than two alleles, and can determine more than two forms of a trait. Multiple alleles may display a dominance hierarchy.
- Alleles that show incomplete dominance are equally dominant. An individual who is heterozygous for alleles that show incomplete dominance will have an intermediate phenotype.
- Codominant alleles are both expressed in a heterozygous individual.

Section 18.4 Questions

1. Multiple alleles control the coat colour of rabbits. A grey colour is produced by the dominant allele *C*. The C^{ch} allele produces a silver-grey colour, called chinchilla, when present in the homozygous condition, $C^{ch}C^{ch}$. When C^{ch} is present with a recessive gene, a light silver-grey colour is produced. The allele C^h is recessive to both the full-colour allele and the chinchilla allele. The C^h allele produces a white colour with black extremities. This coloration pattern is called Himalayan. An allele C^a is recessive to all genes. The C^a allele results in a lack of pigment, called albino. The dominance hierarchy is $C > C^{ch} > C^h > C^a$. **Table 4** provides the possible genotypes and phenotypes for coat colour in rabbits. Notice that four genotypes are possible for full-colour but only one for albino.

Table 4 Coat Colour in Himalayan Rabbits

Phenotypes	Genotypes
full colour	CC, CC^{ch}, CC^h, CC^a
chinchilla	$C^{ch}C^{ch}$
light grey	$C^{ch}C^h$, $C^{ch}C^a$
Himalayan	C^hC^h, C^hC^a
albino	C^aC^a

(a) Indicate the genotypes and phenotypes of the F_1 generation from the mating of a heterozygous Himalayan-coat rabbit with an albino-coat rabbit.
(b) The mating of a full-colour rabbit with a light-grey rabbit produces two full-colour offspring, one light-grey offspring, and one albino offspring. Indicate the genotypes of the parents.
(c) A chinchilla rabbit is mated with a light-grey rabbit. The breeder knows that the light-grey rabbit had an albino mother. Indicate the genotypes and phenotypes of the F_1 generation from this mating.
(d) A test cross is performed with a light-grey rabbit, and the following offspring are noted: five Himalayan rabbits and five light-grey rabbits. Indicate the genotype of the light-grey rabbit.

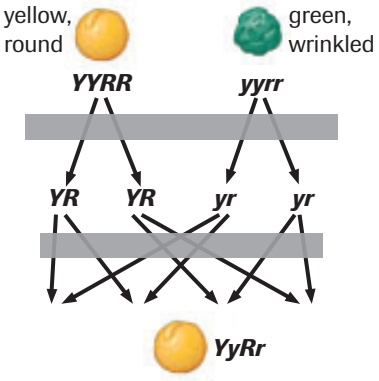
2. A horse that is homozygous for the allele C^r will have a chestnut, or reddish, coat. A horse that is homozygous for the allele C^m will have a very pale cream coat, called cremello. Palomino coat colour is determined by the interaction of both the chestnut and the cremello allele. Indicate the expected genotypic ratio and phenotypic ratio of the F_1 progeny of a palomino horse with a cremello horse.

3. Two pea plants are cross-pollinated. Using a Punnett square and probability analysis, you predict that $\frac{3}{4}$ of the offspring will be tall. However, less than $\frac{1}{4}$ grow to be tall. What other factors can affect phenotype? How much trust should be put on probability calculations?

Dihybrid Crosses and Polygenic Traits 18.5

A **dihybrid cross** is a cross that involves individuals with two independent traits that are present in alternate forms. Mendel performed dihybrid crosses with his garden peas to see if traits were inherited independently or with one another. He first crossed plants that were pure-breeding (homozygous) for two dominant traits with plants that were homozygous for two recessive traits, as shown in **Figure 1**. Each parent is homozygous for two traits, seed shape and seed colour. All the members of the F_1 offspring are heterozygous for the seed-colour gene and for the seed-shape gene. Since all the F_1 progeny had yellow, round seeds, Mendel's principle of dominance applies to this dihybrid cross.

dihybrid cross a genetic cross involving two genes, each of which has more than one allele



All members of the F_1 generation have the same genotype and phenotype.

Figure 1
A dihybrid cross between a pea plant that is homozygous for yellow seed colour (*YY*) and round seed shape (*RR*) with a plant that is homozygous for green seed colour (*yy*) and wrinkled seed shape (*rr*).

Evidence of Independent Assortment

Mendel explained the result shown in **Figure 1** by postulating that each gene was inherited independently. Today, this is referred to as Mendel's second law or the law of *independent assortment*. This law states that genes that are located on different chromosomes assort independently.

To create a Punnett square for a dihybrid cross, we include one allele for both of the genes in the possible gametes. The Punnett square in **Figure 2** shows the expected genotypes and phenotypes for Mendel's dihybrid cross when we assume that the genes for seed shape and seed colour are inherited independently. One parent will produce gametes with alleles *yR* and the other will produce gametes with alleles *Yr*. The predicted phenotype of the F_1 generation is the same as Mendel observed.

Figure 3 shows the behaviour of two separate chromosomes, one that carries the gene for seed shape and another that carries the gene for seed colour. (Pea plants actually have more than two chromosomes.) As the homologous chromosomes move to opposite poles during meiosis, each gamete receives two chromosomes, one carrying the seed-shape gene and one carrying the seed-colour gene. According to the law of segregation, the alleles of both these genes will segregate during meiosis. Therefore, the allele for yellow seeds segregates from the allele for round seeds, and the allele for wrinkled seeds segregates from the allele for round seeds.

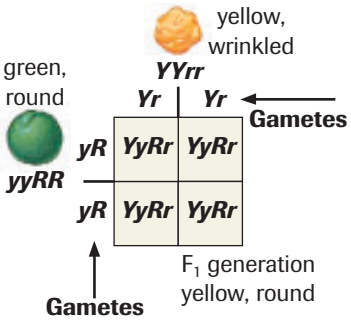


Figure 2
All gametes produced by a pea plant homozygous for yellow seed colour (*YY*) and wrinkled seed shape (*rr*) will have the alleles *Yr*. Similarly, all gametes produced by a pea plant homozygous for green seed colour (*yy*) and round seed shape (*RR*) will have the alleles *yR*. Since all the offspring have yellow, round seeds, the genotype of all the F_1 generation must be *YyRr*. This would not be possible if the genes for seed shape and seed colour were inherited together.

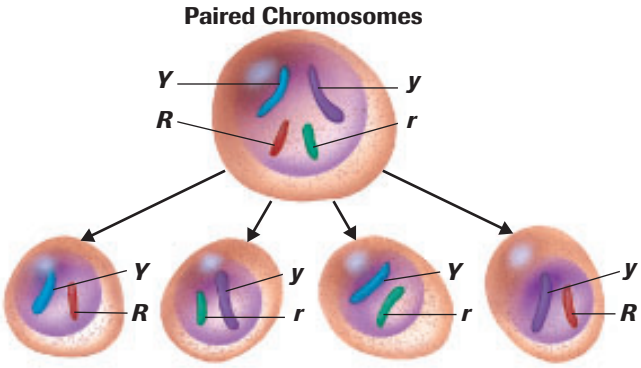


Figure 3
Segregation of alleles and independent assortment of chromosomes during meiosis gives rise to four possible combinations of alleles in the gametes of a plant of genotype *YyRr*.

Mendel then produced an F_2 generation by allowing the F_1 progeny to self-fertilize. He recorded the phenotypes of all the F_2 progeny and then calculated the ratio of each phenotype he observed. The F_2 generation had the following phenotypic ratios: $\frac{9}{16}$ yellow, round seeds; $\frac{3}{16}$ green, round seeds; $\frac{3}{16}$ yellow, wrinkled seeds; and $\frac{1}{16}$ green, wrinkled seeds.

Figure 4 shows the expected genotypes from this cross when we assume that independent assortment occurred. The parents would produce four types of gametes. The genotypes in nine of the 16 cells would determine yellow, round seeds (*YYRR*, *YyRR*, *YYRr*, and *YyRr*); three of the 16 cells would determine green, round seeds (*yyRR* and *yyRr*); three more cells would determine yellow, wrinkled seeds (*YYrr* and *Yyrr*); and one cell would determine green, wrinkled seeds (*yyrr*). Since the predicted phenotypic ratio is the same as the ratio that Mendel observed, this cross also provides evidence for independent assortment.

Gametes	*YR*	*yR*	*Yr*	*yr*
YR	*YYRR*	*YyRR*	*YYRr*	*YyRr*
yR	*YyRR*	*yyRR*	*YyRr*	*yyRr*
Yr	*YYRr*	*YyRr*	*YYrr*	*Yyrr*
yr	*YyRr*	*yyRr*	*Yyrr*	*yyrr*

Figure 4
From the Punnett square analysis, self-fertilization of the F_1 generation will result in an F_2 generation with a 9:3:3:1 ratio. This ratio can only result if segregation of alleles and independent assortment of chromosomes occurs.

INVESTIGATION 18.2 ***Introduction***

Genetics of Corn

Use Punnett squares and phenotypic ratios to analyze the inheritance of two traits in corn.

Report Checklist

○ Purpose	● Design	● Analysis
○ Problem	○ Materials	● Evaluation
○ Hypothesis	○ Procedure	● Synthesis
○ Prediction	● Evidence	

To perform this investigation, turn to page 620.

Probability and Dihybrid Crosses

We can determine the probability of particular phenotypes and genotypes in the progeny of dihybrid crosses in much the same way as for monohybrid crosses. Probability values can be used to predict the chances of getting a particular genotype or phenotype in an offspring, or to tell us whether two genes are likely to be located on different chromosomes. In dihybrid crosses, however, we are interested in finding out the probability that two outcomes will occur at the same time. Recall that probability (P) is given by

$$P = \frac{\text{number of ways that a given outcome can occur}}{\text{total number of possible outcomes}}$$

SAMPLE exercise 1

In humans, free ear lobes are determined by the dominant allele *E*, and attached ear lobes by the recessive allele *e*. The dominant allele *W* determines a widow's peak hairline and the recessive allele *w* determines a straight hairline (**Figure 5**). The genes for these two traits are located on different chromosomes. Suppose a man with the genotype *EeWw* and a woman with the genotype *EeWw* are expecting a child. What is the probability that the child will have a straight hairline and attached ear lobes?

(a)

(b)

(c)

(d)

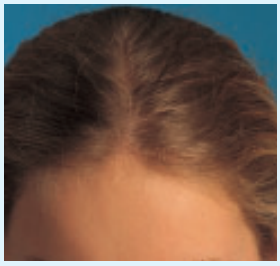


Figure 5
In humans, both ear lobe shape and hairline shape are inherited. The free ear lobe in **(a)** is dominant to the attached ear lobe in **(b)**, and the widow's peak in **(c)** is dominant to a straight hairline in **(d)**.

Solution

To have attached ear lobes and a straight hairline, the child must have the genotype *eeww*. Since the two genes are on separate chromosomes, the gene for ear shape and hairline shape will assort independently. The outcome that the child will receive two *e* alleles is, therefore, independent of the outcome that the child will receive two *w* alleles.

First, determine the probability of each of these outcomes separately, using a separate Punnett square for each gene. From **Figure 6 (a)**, we see the probability that the child will have attached ear lobes is one in four ($\frac{1}{4}$). From **Figure 6 (b)**, we see the probability that the child will have a straight hairline is also one in four ($\frac{1}{4}$).

(a)

	$\frac{1}{2}$ *E*	$\frac{1}{2}$ *e*
$\frac{1}{2}$ *E*	$\frac{1}{4}$ *EE*	$\frac{1}{4}$ *Ee*
$\frac{1}{2}$ *e*	$\frac{1}{4}$ *Ee*	$\frac{1}{4}$ *ee*

(b)

	$\frac{1}{2}$ *W*	$\frac{1}{2}$ *w*
$\frac{1}{2}$ *W*	$\frac{1}{4}$ *WW*	$\frac{1}{4}$ *Ww*
$\frac{1}{2}$ *w*	$\frac{1}{4}$ *Ww*	$\frac{1}{4}$ *ww*

Figure 6
Punnett squares showing monohybrid crosses between heterozygous parents for **(a)** free ear lobes and **(b)** for a widow's peak

Learning Tip

When thinking about probability, keep the following two rules in mind:

- When outcomes are independent, the probability of one outcome is not affected by the result of any other outcomes. For example, if you toss two heads in a row, the probability of tossing heads a third time is still 1 out of 2.
- The probability of independent events occurring together is equal to the *product* of those events occurring separately. The chances of tossing heads once is $\frac{1}{2}$, the probability of tossing heads twice in a row is $\frac{1}{2} \times \frac{1}{2} = \frac{1}{4}$, and the probability of tossing heads three times in a row is $\frac{1}{2} \times \frac{1}{2} \times \frac{1}{2} = \frac{1}{8}$.

EXTENSION

Probability—The Sum and Product Rules

This Audio Clip explores the use of the sum and product rules of probability.

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CAREER CONNECTION

Agrologist

Agrologists are plant, crop, and food production specialists. New breeds of plants and animals are of great interest to these scientists. They work with grain farmers and livestock producers on research projects designed to overcome challenges and realize economic opportunities in agriculture. Learn how agrologists specialize in many fields.

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Now, multiply these probabilities to calculate the probabilities of each event occurring in a dihybrid cross—that is, for the combination of traits. Therefore, the probability that the child will have the genotype *eeww* is $\frac{1}{4} \times \frac{1}{4} = \frac{1}{16}$.

Practice

1. Calculate the probability that the couple will have a child with
   (a) a widow's peak and free ear lobes
   (b) a straight hairline and free ear lobes
   (c) a widow's peak and attached ear lobes

Selective Breeding

selective breeding the crossing of desired traits from plants or animals to produce offspring with both characteristics

The plants and animals that make up the world's food supply have, in large part, been developed artificially from wild ancestors. **Selective breeding** involves identifying individuals with desirable traits and using them as parents for the next generation. Over time, the desirable traits became more and more common. For example, North American Aboriginal farmers used selective breeding to develop many useful crop plants, long before the arrival of Europeans. Many crops that are important to Canadian agriculture were developed by selective breeding, including rust-resistant wheat; sweet, full-kernel corn; and canola, which germinates and grows rapidly in colder climates.

inbreeding the process whereby breeding stock is drawn from a limited number of individuals possessing desirable phenotypes

You are probably familiar with the term "purebreds." Many dogs and horses are considered to be purebreds, or thoroughbreds. Genotypes of these animals are closely regulated by a process called **inbreeding**, in which similar phenotypes are selected for breeding. The desirable traits vary from breed to breed. For example, Irish setters are bred for their long, narrow facial structure and long, wispy hair, but dalmations are bred for broader faces and short hair with spots. The bull terrier (pit bull) was originally bred for fighting. Quick reflexes and strong jaws were chosen as desirable phenotypes. Some geneticists have complained that inbreeding has caused problems for the general public as well as for the breed itself.

DID YOU KNOW

Aboriginal Crop Plants

For centuries, Aboriginal peoples bred many crop plants besides corn, which they ultimately introduced to European settlers. These include beans, tomatoes, potatoes, peanuts, peppers, cocoa, squash, pumpkins, sunflowers, long-fibre cotton, rubber, and quinine.

New varieties of plants and animals can be developed by hybridization. This process is the opposite to that of inbreeding. Rather than breed plants or animals with similar traits, the hybridization technique attempts to blend desirable but different traits. Corn has been hybridized extensively, beginning with the work of Aboriginal peoples thousands of years ago. The hybrids tend to be more vigorous than either parent. **Figure 7**, on the next page, shows the most common method used. Two homozygous plants, A and B, are crossed to produce an AB hybrid. Two other homozygous plants, C and D, are crossed to produce a CD hybrid. Hybrids AB and CD are then crossed to produce hybrid ABCD. This hybrid will have desired traits from plants A, B, C, and D, and will be more vigorous.

Polygenic Traits

polygenic trait inherited characteristics that are determined by more than one gene

In dihybrid crosses, two genes determine two separate traits. However, sometimes a single trait is determined by more than one gene. Many of your characteristics are determined by several pairs of independent genes. Skin colour, eye color, and height are but a few of your characteristics that are **polygenic traits**. Polygenic traits have much more variability in a population than those determined by a single gene. Each of the genes can have multiple alleles, show incomplete dominance or co-dominance, and can be affected by the environment.

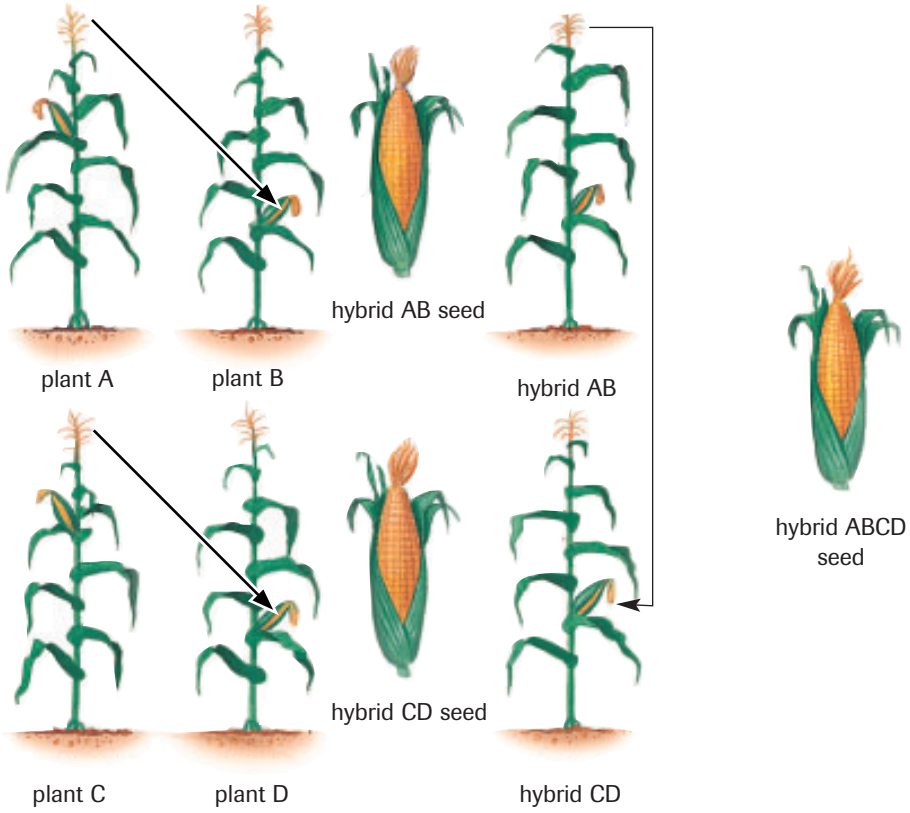

Figure 7
Hybridization can be used to produce a more vigorous strain of corn.

EXTENSION

Coat Colour in Labrador Retrievers

Coat colour variations in this breed of dog is determined by two interacting genes. In this simulation, choose a genotype for each gene and observe the phenotype.

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Examples of polygenic traits in humans include skin colour, height, and intelligence. In other animals and plants, many desirable traits, such as milk production in cows or yield in canola, are also determined by more than one gene pair. This makes breeding for these traits very difficult.

In some cases, two different genotypes interact to produce a phenotype that neither is capable of producing by itself. In other cases, one of the genes will interfere with the expression of the other, masking its effect. Genes that interfere with the expression of other genes are said to be **epistatic**.

Observed phenotypic ratios of polygenic traits vary significantly from the phenotypic ratios predicted by Punnett square analysis of non-interacting genes. Coat colour in dogs provides an examp1e of epistatic genes. As shown in the Punnet square in **Figure 8**, the allele *B* produces black coat-colour, while the recessive allele *b* produces brown coat-colour. However, a second gene also affects coat-colour. The allele *W* of this second gene prevents the formation of pigment, thereby preventing colour. The recessive allele *w* does not prevent colour. The genotype *wwBb* would be black, but the genotype *WwBb* would appear white. The *W* allele masks the effect of the *B* colour gene. In humans, the gene responsible for albinism is epistatic. This gene interferes with the expression of genes that determine pigment formation in the skin, hair, and eyes.

epistatic gene a gene that masks the expression of another gene or genes

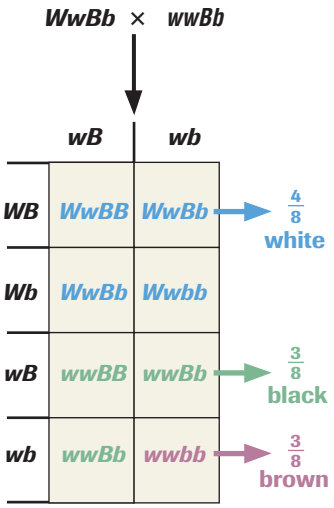

Figure 8
Punnett square of a cross between a white dog (*WwBb*) and a black dog (*wwBb*)

EXPLORE an issue

Issue Checklist

○ Issue	○ Design	● Analysis
● Resolution	● Evidence	● Evaluation

Drought-Tolerant and Salt-Tolerant Plants

Unwise agricultural practices have dramatically reduced the productivity of the world's agricultural land. By one estimate, the reduction in crop yields since 1940 is the same as if all the land in India and China had produced no crops at all. In addition, land equivalent to the area of Hungary has become so degraded that it is unable to produce any viable crop at all. Much of the problem is linked to poor irrigation techniques (**Figure 9**). When water, rich in minerals, floods the land, evaporation carries away water but leaves the minerals. Eventually, the mineral salts accumulate within the soil. creating an environment difficult for plants to survive.

Proposed Solutions from Genetics

Traditionally, plant breeders have used selective breeding to create new varieties with desirable traits. Today, molecular biologists have developed gene insertion techniques that provide breeders with a more precise tool. Using gene splicing, desired traits from one species can be introduced into a non-related species.

In 2001, articles in scientific journals reported the production of genetically modified (GM) tomatoes that can grow in soils with high salt levels. Researchers inserted a gene that enhanced the ability of cells in the tomato plants to transport excess salts into fluid storage sacs (vacuoles). The GM tomatoes can grow in soils 50 times more saline than non-GM tomatoes. The salts accumulate in the leaves, so the tomato fruit does not have a salty taste. The development of other plants capable of living in saline solutions will allow farmers to reclaim marginal land.

In related research, geneticists are looking at developing drought-tolerant plants. Several genes have been identified that enable plants to cope with arid conditions. The Rockefeller Foundation committed $50 million to support the effort to improve drought resistance for GM maize and rice.

However, as with any technology, GM drought-tolerant and salt-tolerant plants could have undesirable consequences. Some of these concerns are outlined below.

Environmental Concerns: Every year, some of the best farmland in the world is converted to urban land. This expansion of cities into farmland also reduces food production. Producing GM drought-tolerant and salt-tolerant plants that can grow on marginal land does nothing to resolve the issue of urban expansion.

GM drought-tolerant and salt-tolerant plants could lead to the conversion of deserts and saltwater marshes into agricultural land, disrupting the natural balance within these ecosystems. These ecosystems provide habitat for many species, and saltwater marshes also help filter and clean water systems.

Food Production Concerns: At present, 5 billion people inhabit Earth, and the population is projected to increase to nearly 10 billion within 50 years. Only 3.7 billion ha (hectares) of the world's 13.1 billion ha of land can be used for crop production. According to the United Nations Food and

Figure 9
Irrigation allows plants to grow in arid lands.

Agricultural committee, over the next 50 years, the amount of arable land on Earth per person will decline from 0.24 ha to about 0.12 ha, which will not be enough to feed many of the poor. Although GM crops may not be the entire answer, they may allow an increase in food production, and so deserve further study.

Geneticists' Concerns: Some geneticists worry about the consequences if GM crops hybridize with non-GM species. Traditional methods of crop breeding involve selecting particular individuals with desirable traits from within a population, thereby altering gene frequencies within a population of a single species. Newer technologies allow genes to be transferred between entirely different species. It is difficult to predict how these transferred genes will interact in a naturally reproducing population. For example, would a gene that increases drought tolerance also make a plant more susceptible to disease?

- Evaluate each of the concerns expressed.
  - (a) What assumptions lie at the basis of these divergent opinions?
  - (b) What additional information would be useful to make an informed decision about whether or not GM crops should be pursued?
- Working in a group, discuss the different viewpoints presented above.
- Still in your group, conduct additional research on the issue of developing GM drought-tolerant and salt-tolerant plants. When research is complete, discuss the question below until you reach a consensus.
  - (c) Should GM crops, resistant to drought and salinity, be funded? Do they provide at least a partial solution?
- Be prepared to debate the issue as a class. Express your opinion and provide a rationale for your view.

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SUMMARY Dihybrid Crosses

- The phenotypic ratios that Mendel observed in his dihybrid crosses provide evidence for independent assortment of chromosomes.
- The probability of inheritance of the two traits together is the same as the product of the probability of inheritance of both traits separately.

Section 18.5 Questions

1. In guinea pigs, black coat colour (*B*) is dominant to white (*b*), and short hair length (*S*) is dominant to long (*s*). Indicate the genotypes and phenotypes from the following crosses:
   (a) A guinea pig that is homozygous for black and heterozygous for short hair crossed with a white, long-haired guinea pig.
   (b) A guinea pig that is heterozygous for black and for short hair crossed with a white, long-haired guinea pig.
   (c) A guinea pig that is homozygous for black and for long hair crossed with a guinea pig that is heterozygous for black and for short hair.

2. Black coat colour (*B*) in cocker spaniels is dominant to white coat colour (*b*). Solid coat pattern (*S*) is dominant to spotted pattern (*s*). The gene for pattern arrangement is located on a different chromosome than the one for colour, and the pattern gene segregates independently of the colour gene. A male that is black with a solid pattern mates with three females. The mating with female A, which is white and solid, produces four pups: two black, solid, and two white, solid. The mating with female B, which is black and solid, produces a single pup, which is white, spotted. The mating with female C, which is white and spotted, produces four pups: one white, solid; one white, spotted; one black, solid; one black, spotted. Indicate the genotypes of the parents.

3. For human blood, the alleles for types A and B are codominant, but both are dominant over the type O allele. The Rh factor is separate from the ABO blood group and is located on a separate chromosome. The Rh+ allele is dominant to Rh−. Indicate the possible phenotypes of a child of a woman with type O, Rh− and a man with type A, Rh+.

4. Skin colour in humans is determined by more than one gene pair, whereas Rh factor in blood is controlled by one gene pair. Which would show more variability in the human population? Why?

INVESTIGATION 18.1

How Do Environmental Factors Affect Gene Expression?

Many environmental factors can affect the phenotype of a plant. Traits such as growth rate, colour, leaf size, and leaf shape can be affected by environmental factors such as light intensity, hours of darkness, wavelength of radiation, and air temperature. In this investigation, you will design an experiment to explore how one environmental factor of your choice affects the phenotype of a plant.

Report Checklist

● Purpose	● Design	● Analysis
● Problem	● Materials	● Evaluation
● Hypothesis	● Procedure	○ Synthesis
● Prediction	● Evidence	

You can find more information about designing an experiment in Appendix A1. Have your teacher check the procedure before beginning the experiment. Then, write a lab report, following the guidelines in Appendix A3.

INVESTIGATION 18.2

Genetics of Corn

Corn is one of the world's most important food crops. It has been subject to selective breeding techniques and hybridization for many years, which have resulted in vigorous, high-yielding varieties. Nearly all corn grown today is hybrid corn. Some varieties of corn are chosen for their sweet flavour while the mixed coloration of other, inedible varieties makes them popular decorations during the autumn months.

Report Checklist

○ Purpose	● Design	● Analysis
○ Problem	○ Materials	● Evaluation
○ Hypothesis	○ Procedure	● Synthesis
○ Prediction	● Evidence	

Purpose

To determine the genotypes of parents by examining phenotypes of corn for two different and independent traits.

Problem

To determine the probable genotypes of the parents of the sample corn ears.

Materials

dihybrid corn ears (sample A, sample B)

Procedure

1. Obtain a sample A corn ear from your instructor (**Figure 1**). The kernels display two different traits that are located on different chromosomes.

Figure 1

(a) Indicate the two different traits.

(b) Predict the dominant phenotypes.

(c) Predict the recessive phenotypes.

INVESTIGATION 18.2 *continued*

2. Assume that the ear of corn is from the F_2 generation. The original parents were pure breeding homozygous for each of the characteristics. Assign the letters *P* and *p* to the alleles for colour, and *S* and *s* to the alleles for shape. Use the symbols *PPss* × *ppSS* for the parent generation.

(d) Indicate the phenotype of the *PPss* parent.

(e) Indicate the phenotype of the *ppSS* parent.

3. Count 100 of the kernels in sequence, and record the actual phenotypes in a table similar to **Table 1**.

Table 1 Phenotypes of the F_2 Generation

Phenotype	Number	Ratio
dominant genes for colour and shape		
dominant gene for colour, but recessive for shape		
recessive gene for colour, but dominant gene for shape		
recessive genes for colour and shape		

4. Obtain sample B. Assume that this ear was produced from a test cross. Count 100 kernels in sequence and record your results.

Analysis and Evaluation

(f) Indicate the expected genotypes and phenotypes of the F_1 generation resulting from a cross between the original parents *PPss* × *ppSS*.

(g) Use a Punnett square to show the expected genotypes and the phenotypic ratio of the F_2 generation. Compare your results with what you obtained in question 3. What factors might account for discrepancies?

(h) Assuming that sample B was produced from a test cross, indicate the phenotypic ratio of the F_1 generation.

(i) Indicate the phenotype of the unknown parent.

Synthesis

(j) Why are test crosses important to plant breeders?

(k) A dihybrid cross can produce 16 different combinations of alleles. Explain why 100 seeds were counted rather than only 16.

(l) A dominant allele *Su*, called starchy, produces smooth kernels of corn. The recessive allele *su*, called sweet, produces wrinkled kernels of corn. The dominant allele *P* produces purple kernels, while the recessive *p* allele produces yellow kernels. A corn plant with starchy, yellow kernels is cross-pollinated with a corn plant with sweet, purple kernels. One hundred kernels from the hybrid are counted, and the following results are obtained: 52 starchy, yellow kernels and 48 starchy, purple kernels. What are the genotypes of the parents and the F_1 generation?

(m) The wild ancestor of corn grew only in Central America. From this ancestor, Aboriginal peoples used selective breeding to develop different types of corn. Today, scientists continue to use technology and selective breeding methods to develop varieties of corn that can grow in a wide range of environmental conditions. As a result, corn is now grown in many places where its ancestor would not be able to survive. What are some risks associated with growing a species in a foreign environment?

EXTENSION

Comb Shape in Chickens

Two genes interact to produce comb shape in chickens. Change the genotype and see what happens to the phenotype .

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Chapter 18 SUMMARY

Outcomes

Knowledge

- describe the evidence for dominance, segregation, and the independent assortment of genes on different chromosomes, as investigated by Mendel (18.1, 18.2)
- compare ratios and probabilities of genotypes and phenotypes for dominant/recessive alleles, multiple alleles, and incompletely dominant or codominant alleles, epistatic, and pleiotropic alleles (18.2, 18.3, 18.4, 18.5)
- explain the relationship between variability and the number of genes controlling a trait (18.3)

STS

- explain that decisions regarding the application of scientific and technological development involve a variety of perspectives (18.3)

Skills

- ask questions and plan investigations by designing a plan for collecting data to demonstrate human inheritance (18.2)
- conduct investigations and gather and record data by performing an experiment to demonstrate inheritance of a trait controlled by a single pair of genes (18.5), and by designing and performing an experiment to demonstrate that an environmental factor can cause a change in the expression of genetic information in an organism (18.4)
- analyze data and apply mathematical and conceptual models by predicting, quantitatively, the probability of inheritance from monohybrid and dihybrid (18.2, 18.4); using Punnett squares to interpret patterns and trends associated with monohybrid and dihybrid patterns of inheritance (18.2, 18.4); performing, recording, and explaining predicted phenotypic ratios versus actual counts in genetic crosses to show a relationship between chance and genetic results (18.2, 18.4, 18.5); and drawing and interpreting pedigree charts from data on human single-allele and multiple-allele inheritance patterns (18.3, 18.4)
- work as members of a team and apply the skills and conventions of science (all)

Key Terms

18.1

progeny
dominant trait
recessive trait
allele
homozygous
heterozygous
genotype
phenotype
segregation

18.2

phenotypic ratio
Punnett square
genotypic ratio
test cross

18.3

pedigree chart

18.4

pleiotropic gene
wild type
mutant
incomplete dominance
codominance

18.5

dihybrid cross
selective breeding
inbreeding
polygenic trait
epistatic gene

MAKE a summary

1. Create a concept map that shows the principles of inheritance of traits. Label the sketch with as many of the key terms as possible.
2. Revisit your answers to the Starting Points questions at the start of the chapter. Would you answer the questions differently now? Why?

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The following components are available on the Nelson Web site. Follow the links for *Nelson Biology Alberta 20–30*.

- an interactive Self Quiz for Chapter 18
- additional Diploma Exam-style Review Questions
- Illustrated Glossary
- additional IB-related material

There is more information on the Web site wherever you see the Go icon in the chapter.

EXTENSION

CBC radioONE QUIRKS & QUARKS

Spawning Trouble

Dr. Daniel Heath, (University of Windsor) has discovered that the eggs of captive-bred salmon are getting smaller each year. The lack of selective pressure on the eggs in a hatchery may be the cause, since more small fish are surviving than would be if the eggs developed in the wild. Dr. Heath is concerned this will lead to health problems in the wild population, and if this may also be a general problem with captive breeding programs for other animals, including endangered species.

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Many of these questions are in the style of the Diploma Exam. You will find guidance for writing Diploma Exams in Appendix A5. Science Directing Words used in Diploma Exams are in bold type. Exam study tips and test-taking suggestions are on the Nelson Web site.

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DO NOT WRITE IN THIS TEXTBOOK.

Part 1

Use the following information to answer questions 1 and 2.

Long stems are dominant over short stems for pea plants. A heterozygous long-stem plant is crossed with a short-stem plant.

1. Determine and identify the genotypic ratio of the F_1 progeny from the cross.
   A. 50 % *Ss* and 50 % *ss*
   B. 75 % *SS* and 25 % *Ss*
   C. 75 % *Ss* and 25 % *ss*
   D. 100 % *Ss*

2. Determine and identify the phenotypic ratios of the F_1 progeny of the cross.
   A. 75 % long stem and 25 % short stem
   B. 50 % long stem and 50 % short stem
   C. 75 % short stem and 25 % long stem
   D. 100 % long stem

Use the following information to answer questions 3 to 5.

The pedigree chart in **Figure 1** shows the transmission of blood types in a family.

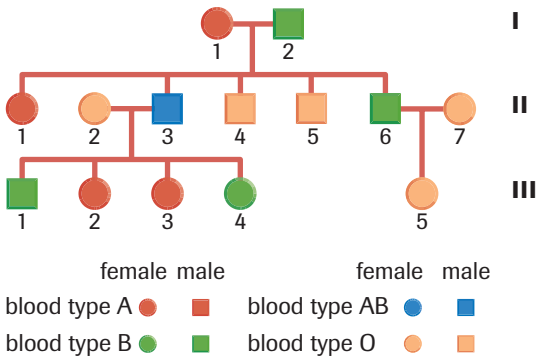


Figure 1

3. Indicate the genotypes for individuals 1 and 2, generation I.
   A. I^Ai and I^Bi
   B. I^AI^A and I^BI^B
   C. I^Ai and I^BI^B
   D. I^AI^B and I^Bi

4. NR Predict the chance of parents 1 and 2 from generation I having a child with blood type AB. (Record your answer in decimal form.)

5. NR If individuals 6 and 7 had another child, calculate the probability that the child would have blood type O. (Record your answer in decimal form.)

Use the following information to answer questions 6 and 7.

In cattle, the polled trait (hornless) is dominant to the horned condition. A single bull mates with three different cows and produces offspring as shown in **Figure 2**.

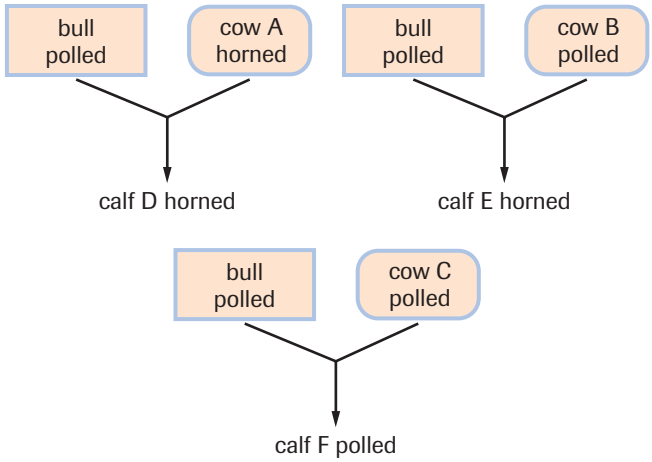


Figure 2

6. Identify the respective genotypes for the bull, cow A, and cow B.
   A. bull = *Pp*, cow A = *pp*, cow B = *Pp*
   B. bull = *PP*, cow A = *pp*, cow B = *Pp*
   C. bull = *Pp*, cow A = *pp*, cow B = *pp*
   D. bull = *PP*, cow A = *Pp*, cow B = *Pp*

7. Identify which of the cattle could have two possible genotypes.
   A. cow C and calf F
   B. cow B and calf E
   C. cow A and calf D
   D. bull and calf D

Part 2

8. **Explain** the advantages and limitations of using blood typing by the courts to prove paternity.

9. Cystic fibrosis is regulated by a recessive allele, *c*. **Explain** how two parents without this condition can produce a child with cystic fibrosis.

10. In horses, the trotter trait is dominant to the pacer trait. A male, described as a trotter, mates with three different females. Each female produces a foal. The first female, a pacer, gives birth to a foal that is a pacer. The second female, also a pacer, gives birth to a foal that is a trotter. The third female, a trotter, gives birth to a foal that is a pacer. **Determine** the genotypes of the male, all three females, and the three foals sired. Designate the trotter allele as *T* and the pacer allele as *t*.

11. For ABO blood groups in humans, the A and B genes are codominant, but both A and B are dominant over type O.
(a) **Identify** the possible blood types in the children of a man with blood type O and a woman with blood type AB.
(b) Could a woman with blood type AB ever produce a child with blood type AB? Could she ever have a child with blood type O? **Explain** your answer.

12. Some cats have six toes, a condition determined by a dominant allele. **Sketch** a pedigree chart showing the mating of a male cat with six toes to a normal female. Assume the following:
- The male cat with six toes had a normal mother.
- The cats produce six offspring (four females and two males). Two of the female offspring and one of the male offspring have six toes.
- One of the six-toed female offspring mates with a six-toed male from different parents. Four female offspring are produced, and three of them have six toes.

13. In shorthorn cattle, the mating of a red bull and a white cow produces a calf that is described as roan. Roan animals have intermingled red and white hair. After many matings between roan bulls and roan cows, the following phenotypic ratio was observed in the offspring: one red, two roan, one white. Does this ratio indicate codominance or multiple alleles? **Explain** your answer.

Use the following information to answer questions 14 to 16.

Thalassemia is a serious human genetic disorder which causes severe anemia. The homozygous condition (T^mT^m) leads to severe anemia. People with thalassemia die before sexual maturity. The heterozygous condition (T^mT^n) causes a less serious form of anemia. The genotype T^nT^n causes no symptoms of the disease.

DE **Predict** all the possible genotypes of the offspring of a male with the genotype T^mT^n and a woman of the same genotype.

DE **Predict** all the possible phenotypes of the offspring of a man with the genotype T^mT^n and a woman of the same genotype.

DE Would it ever be possible for offspring to be produced from two individuals with the genotypes T^mT^m and T^mT^n respectively? **Explain** your answer.

Use the following information to answer questions 17 and 18.

Baldness is an autosomal trait, but it is influenced by sex. Baldness (*HB*) is dominant in males but recessive in females. The normal gene (*Hn*) is dominant in females, but recessive in males.

17. DE **Explain** how a bald offspring can be produced from the mating of a normal female and a normal male.

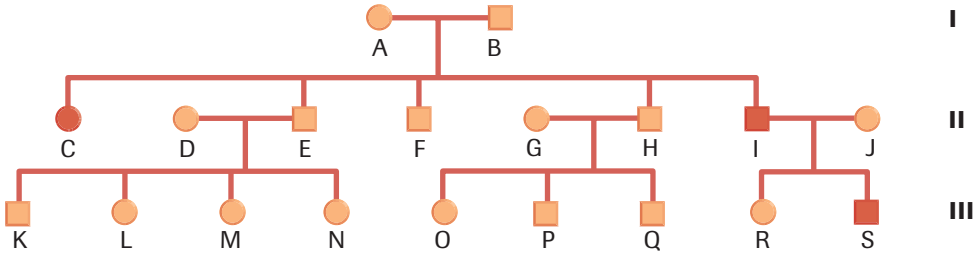
18. DE Could normal parents ever produce a bald girl? **Explain** your answer.

19. The ability to curl your tongue up on the sides (*T*) is dominant to not being able to roll your tongue (*t*).
(a) A woman who can roll her tongue marries a man who cannot. Their first child has his father's phenotype. **Predict** the genotypes of the mother, father, and child.
(b) **Determine** the probability that their second child will not be able to roll her or his tongue.

20. Phenylketonuria (PKU) is an inherited disease caused by the lack of the enzyme needed to metabolize the amino acid phenylalanine. If untreated, PKU builds up in the brain and causes mental retardation. PKU is determined by a recessive allele. A woman and her husband are both carriers of PKU. **Determine** the probability of
(a) their first child having PKU.
(b) both of their first two children having PKU.

21. DE Amniocentesis is a common prenatal procedure, used to obtain cells to test for genetic abnormalities such as cystic fibrosis. The test is usually carried out in the 15th to 18th week of pregnancy when a woman has an increased risk of having children with genetic abnormalities. A woman with cystic fibrosis in her family history (**Figure 3**, next page) is carrying a child. Her husband's lineage also is linked to cystic fibrosis. Cystic fibrosis is caused by a recessive allele found on chromosome 7. Write a unified response addressing the following aspects of performing amniocentesis in the case of father K and mother O.
- Like all procedures that enter the body, some risk, although small, is associated with amniocentesis. On the basis of the information provided, would you recommend an amniocentesis be done for mother O and father K? **Explain** your reasons.
- Would you recommend the procedure if father K had married mother O's cousin, woman J? **Explain** your reasons.
- Should amniocentesis be performed even if there is no strong evidence suggesting genetic problems? **Explain** your reasons.
- Should this pedigree be made public? **Identify** both pros and cons before coming to a conclusion.

Father K's Family Tree



Mother O's Family Tree

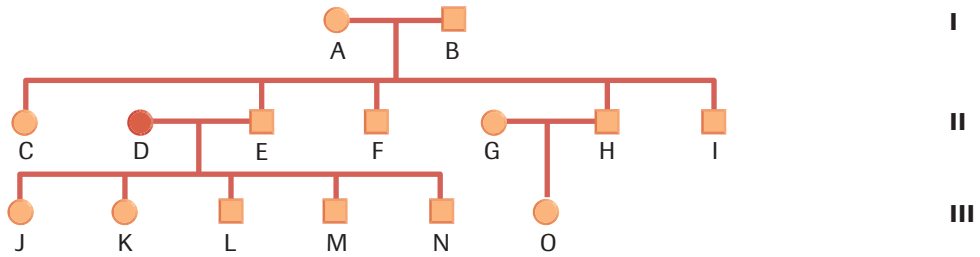


Figure 3

22. In Canada, it is illegal to marry your immediate relatives. Using the principles of genetics, **explain** why inbreeding of humans is discouraged.

Use the following information to answer questions 23 to 26.

When paper impregnated with the bitter chemical phenylthiocarbamide (PTC) is placed on the tongue, about 70 % of people can taste the chemical. The ability to taste PTC is determined by a dominant taster allele (*T*). Those who cannot taste PTC are homozygous for the recessive alleles (*t*). A second gene on another chromosome determines skin pigmentation. Allele (*A*) is dominant, and determines normal pigmentation. People who are homozygous for the recessive allele (*a*) will be albino. A normally pigmented woman who cannot taste PTC has a father who is an albino and a PTC taster. She marries a normally pigmented man who is homozygous for the dominant (A) allele for pigmentation. The man can taste PTC, but his mother cannot.

23. **Predict** all the possible genotypes for these two traits for children by this couple. DE

24. **Determine** the probability that a child from this couple will not be able to taste PTC. DE

25. **Determine** the probability that a child from this couple will be albino? DE

26. **Determine** the probability that a child from this couple will be able to taste PTC and be albino. DE

Use the following information to answer questions 27 to 29.

In a specific variety of soybeans, the allele for seeds containing a high oil-content (*H*) is dominant to the allele for low oil-content (*h*). A gene located on another chromosome determines the number of seeds in a pod. Through crossing experiments, it was determined that the allele that determines four seeds per pod (*E*) is dominant to the allele that determines two seeds per pod (*ee*). A plant breeder crosses two soybean plants of this variety, both of which have high oil-content and four seeds per pod. The phenotypes of the F_1 generation and their ratios are shown in **Table 1**.

Table 1 Phenotypes of the F_1 Generation

Phenotype	Ratio
high oil-content—four seeds per pod	9
high oil-content—two seeds per pod	3
low oil-content—four seeds per pod	3
low oil-content—two seeds per pod	1

27. **Predict** the genotypes of the parent plants. DE

28. The plant breeder crosses two individuals from the F_1 generation that have high oil-content and four seeds per pod. If all the members of the F_2 generation all have high oil-content and four seeds per pod, **predict** the genotypes of the two F_1 parent plants chosen by the breeder. DE

29. The breeder wants to confirm the genotype of the two F_1 parent plants using a cross. What genotype should the plant she crosses the F_1 parent plants have? **Explain**. DE

chapter

19 Beyond Mendel

In this chapter

- Exploration: Inherited Traits
- Lab Exercise 19.A: Tracing the Hemophilia Gene
- Explore an Issue: Screening for Genetic Disorders
- Web Activity: Amniocentesis
- Investigation 19.1: Sex-Linked Traits
- Lab Exercise 19.B: Mapping Chromosomes
- Lab Exercise 19.C: Evidence of Hereditary Material
- Web Activity: Avery and MacLeod
- Web Activity: Elementary, My Dear Crick
- Investigation 19.2: Isolation and Quantification of DNA
- Explore an Issue: Competition and Collaboration Advance Science

Early scientists believed that hereditary traits were located in the blood. The term "pure bloodline," which is still used today by animal breeders (**Figure 1**), is a reminder of this misconception, as is the French term Métis conferred by European fur traders on peoples of mixed Aboriginal and European "blood." Today we know that inherited traits are determined by genes, which are located along the thread-like chromosomes found in the nucleus of each cell.

The field of genetics changed quickly once scientists began to describe the location and the chemical makeup of chromosomes. Genes can now be identified and selected, and sometimes even altered. One of the most dramatic examples of changing inherited traits is the production of mice that are smarter than mice are naturally. This genetically modified strain was dubbed Doogie, after a television character who was a teenage genius.

The modification and insertion of a single gene, *NR2B*, into a chromosome of the mice improves the functioning of nerve receptors that play a key role in memory and learning. The laboratory-bred Doogie mice learn faster and remember more than normal mice. For example, scientists found that when a new and an old object were introduced into the cage with the Doogie mice, they spent most of their time exploring the new object. This indicated that they recognized and remembered the old object. Normal mice spent equal time with the new and old objects. The Doogie mice generated great excitement, because humans possess a corresponding gene.

STARTING Points

Answer these questions as best you can with your current knowledge. Then, using the concepts and skills you have learned, you will revise your answers at the end of the chapter.

1. In what part of the cell would you find genes?
2. Can you distinguish males from females by looking at their genetic material?
3. Explain how a better understanding of chromosome structure could lead to a more complete understanding of gene function.
4. Why might some people be opposed to making mice smarter?
5. Why might the research with mice prove important for people with Alzheimer's disease?

Career Connection: Entomologist

Figure 1
Animal breeders produce varieties of a species with a specific set of traits, such as these Appaloosa horses. The value of an individual animal is often determined by its bloodline, a term that dates back to early misconceptions about heredity.

Exploration Inherited Traits

Some physical characteristics are controlled by a single gene that can be expressed in one of two ways. Try the tests below to see what phenotype you express.

- Fold your arms in front of your body.
  (a) Which arm is on top?
- Change arm position so that the other arm is on top.
  (b) Describe how it feels.
- Interlock your fingers.
  (c) Are the fingers from your left hand or your right hand on top?
- Place a strip of PTC paper on your tongue.
  (d) Could you taste the paper?
- Gather and compile the class data for all three tests.
  (e) For each test, which trait occurred most frequently in your class?
  (f) Do traits determined by dominant genes always occur with the highest frequency? Explain your answer.

19.1 Chromosomes and Genetics

Figure 1
The artist Leonardo da Vinci became interested in anatomy and dissection because of his desire to paint the human form better.

During the Middle Ages (500–1300 CE), curious individuals would sneak into caves to dissect corpses. Despite strict laws prohibiting such behaviour, the inquiring minds of early physicians and scientists compelled them to conduct their investigations. Generations of artists sketched different parts of the body (**Figure 1**), creating a guide to anatomy in the process. As a composite structure of organs began to appear, theories about function arose. The principle that structure gives clues about function also applies to genetics. However, the early geneticists had to wait for the emergence of the light microscope before investigations into genetic structure could seriously progress. The study of genes is closely connected with technology. The light microscope, the electron microscope, X-ray diffraction, and gel electrophoresis have provided a more complete picture of the mechanisms of gene action.

The discovery of the nucleus in 1831 was an important step toward understanding the structure and function of cells and the genes they contain. By 1865, the year in which Mendel published his papers, biologists knew that the egg and sperm unite to form a zygote, and it was generally accepted that factors from the egg and sperm were blended in developing the characteristics of the offspring. Even though Mendel knew nothing about meiosis or the structure or location of the hereditary material, he was able to develop theories about inheritance that adequately explain how traits are passed on from generation to generation.

At about the same time that Mendel was conducting his experiments with garden peas, new techniques in lens grinding were providing better microscopes. The improved technology helped a new branch of biology, cytology, to flourish. Cytology is the study of cell formation, structure, and function. Aided by these technological innovations, in 1882, Walter Fleming described the separation of threads within the nucleus during cell division. He called the process mitosis. In the same year, Edouard van Benden noticed that the sperm and egg cells of roundworms had two chromosomes, but the fertilized eggs had four chromosomes. By 1887, August Weisman offered the theory that a special division took place in sex cells. By explaining the reduction division now known as meiosis, Weisman added an important piece to the puzzle of heredity and provided a framework in which Mendel's work could be understood. When scientists rediscovered Mendel's experiments in 1900, the true significance of his work became apparent.

Chromosomal Theory

Learning Tip

Recall that homologous chromosomes occur in pairs and are similar in size, shape, and gene information and arrangement.

In 1902, American biologist Walter S. Sutton and German biologist Theodor Boveri independently observed that chromosomes came in pairs that segregated during meiosis. The chromosomes then formed new pairs when the egg and sperm united. The concept of paired, or homologous, chromosomes supported Mendel's explanation of inheritance based on paired factors. Today, these factors are referred to as the alleles of a gene. One factor, or allele, for each gene comes from each sex cell.

The union of two different alleles in offspring and the formation of new combinations of alleles in succeeding generations could be explained and supported by cellular evidence. The behaviour of chromosomes during gamete formation could help explain Mendel's law of segregation and law of independent assortment.

Sutton and Boveri knew that the expression of a trait, such as eye colour, was not tied to only the male or only the female sex cell. Some structures in both the sperm cell and

the egg cell must determine heredity. Sutton and Boveri deduced that Mendel's factors (alleles) must be located on the chromosomes. The fact that humans have 46 chromosomes (44 **autosomes** and 2 sex chromosomes), but thousands of different traits, led Sutton to hypothesize that each chromosome carries genes. Genes that are on the same chromosome are said to be **linked genes**.

autosome a chromosome not involved in sex determination

linked genes genes that are located on the same chromosome

The chromosomal theory of inheritance can be summarized as follows:

- Chromosomes carry genes, the units of heredity.
- Paired chromosomes segregate during meiosis. Each sex cell or gamete has half the number of chromosomes found in the somatic cells. This explains why each gamete has only one of each of the paired alleles.

As you saw in the previous chapter, chromosomes assort independently during meiosis. Each gamete receives one member from each pair of chromosomes, and each chromosome pair has no influence on the movement of any other chromosome pair. This explains why in a dihybrid cross an F_1 parent, *AaBb*, produces four types of gametes: *AB*, *aB*, *Ab*, *ab*. Each gamete appears with equal frequency due to segregation and independent assortment. Each chromosome contains many different alleles and each gene occupies a specific locus or position on a particular chromosome.

Morgan's Experiments and Sex-Linked Traits

The American Thomas Hunt Morgan was among the first of many geneticists who used the tiny fruit fly, *Drosophila melanogaster*, to study the principles of inheritance. There are several reasons why the fruit fly is an ideal subject for study. First, the fruit fly reproduces rapidly. Offspring are capable of mating shortly after leaving the egg, and females produce over 100 eggs after each mating. Female *Drosophila* can reproduce for the first time when they are only 10 to 15 days old, so it is possible to study many generations in a short period of time. Since genetics is based on probability, the large number of offspring is ideal. A second benefit arises from *Drosophila*'s small size. Many individuals can be housed in a single culture tube. A small, solid nutrient at the bottom of the test tube can maintain an entire community. The third and most important quality of *Drosophila* is that males can easily be distinguished from females. Males are smaller and have a rounded abdomen with a dark-coloured posterior segment while the larger females have a pointed abdomen with a pattern of dark bands.

Figure 2
In *Drosophila*, the allele that codes for white eyes (male fly, top photo) is recessive to the allele that codes for red eyes (female fly, bottom photo).

While examining the eye colour of a large number of *Drosophila*, Morgan noted the appearance of a white-eyed male among many red-eyed offspring (**Figure 2**). He concluded that the white-eyed trait must be a mutation. Morgan was interested in tracing the inheritance of the allele coding for white eyes, so he mated the white-eyed male with a red-eyed female. All members of the F_1 generation had red eyes. Normal Mendelian genetics indicated that the allele for red eyes was dominant. Most researchers might have stopped at that point, but Morgan did not. Pursuing further crosses and possibilities, he decided to mate two hybrids from the F_1 generation. An F_2 generation produced $\frac{3}{4}$ red eyes and $\frac{1}{4}$ white eyes, a ratio that could again be explained by Mendelian genetics. But further examination revealed that all the females had red eyes. Only the males had white eyes. Half of the males had red eyes and half had white eyes. Did this mean that the white-eyed phenotype only appears in males? Why could males express the white-eyed trait but not females? How did the pattern of inheritance differ between males and females? To find an answer, Morgan turned to cytology.

Previous researchers had stained and microscopically examined the eight chromosomes from the cells of the salivary glands of *Drosophila*. They found that females have four homologous pairs and males have only three homologous pairs. The fourth pair, which determines sex, is only partially homologous. Males were found to have one

CAREER CONNECTION

Entomologist

Entomologists study the life cycle of insects and conduct research into evolution and biodiversity. The science of entomology has made a significant contribution to understanding genetics and gene mapping. Would you like to work with fruit flies or arthropods, such as spiders and mites? Explore this field of study.

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X chromosome paired with a small, hook-shaped Y chromosome. Females have two paired X chromosomes (**Figure 3**). Since the X and Y chromosomes are not completely homologous (although they act as homologous pairs during meiosis), it was concluded that they contain different genes.

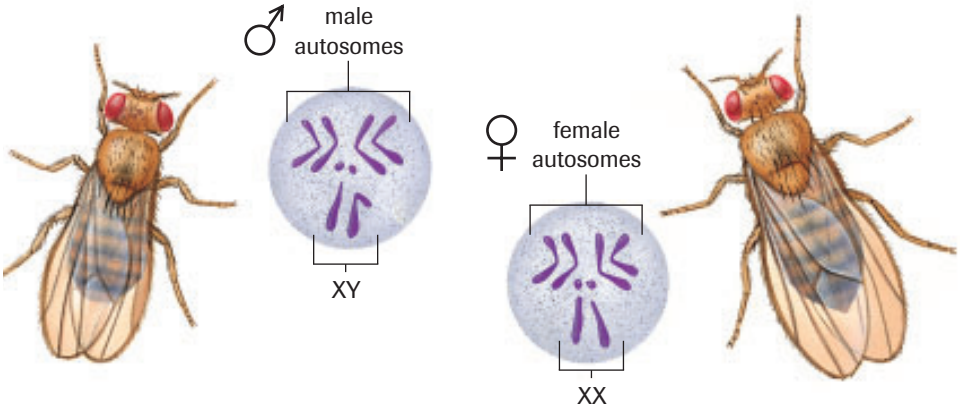

Figure 3
Drosophila contain three pairs of autosomes and a single pair of sex chromosomes.

Morgan explained the results of his experiments by concluding that the Y chromosome does not carry the gene to determine eye colour. We now know that the gene for eye colour in *Drosophila* is located on the part of the X chromosome that does not match the Y chromosome. Therefore, Morgan's conclusion was correct. The Y chromosome does not carry an allele for the eye-colour gene. Traits determined by genes located on sex chromosomes are called **sex-linked traits**.

sex-linked trait trait that is determined by genes located on the sex chromosomes

The initial problem can now be re-examined. The pure-breeding, red-eyed female can be indicated by the genotype X^RX^R and the white-eyed male by the genotype X^rY. The symbol X^R indicates that the allele for red eye is dominant and is located on the X chromosome. There is no symbol for eye colour on the Y chromosome because it does not contain an allele for the trait. A Punnett square, as shown in **Figure 4**, can be used to

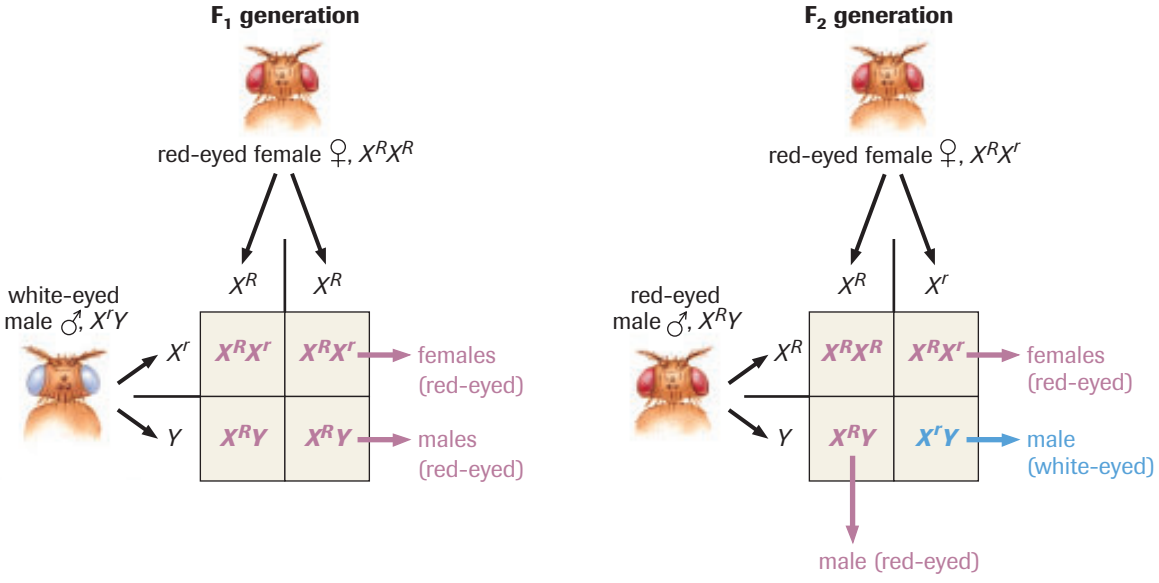

Figure 4
Punnett squares showing F_1 and F_2 generations for a cross between a homozygous red-eyed female and a white-eyed male.

determine the genotypes and the phenotypes of the offspring. All members of the F_1 generation have red eyes. The females have the genotype X^RX^r, and the males have the genotype X^RY.

The F_2 generation is determined by a cross between a male and female from the F_1 generation. Upon examination of the F_1 and F_2 generations, the question arises whether the males inherit the trait for eye colour from the mother or father. The male offspring always inherit a sex-linked trait from the mother. The father supplies the Y chromosome, which makes the offspring male.

The F_2 male *Drosophila* are X^RY and X^rY. The females are either homozygous red for eye colour, X^RX^R, or heterozygous red for eye colour, X^RX^r (**Table 1**). Although Morgan did not find any white-eyed females from his initial cross, some white-eyed females do occur in nature. For this to happen, a female with at least one allele for white eyes must be crossed with a white-eyed male. Notice that females have three possible genotypes, but males have only two. Males cannot be homozygous for an X-linked gene because they have only one X chromosome. The Y chromosome has less than 100 genes.

Recall that humans have 46 chromosomes. Females have 23 pairs of homologous chromosomes: 22 autosomes, and two X sex chromosomes. Males have 22 pairs of homologous chromosomes, and one X sex chromosome and one Y sex chromosome (**Figure 5**). It has been estimated that the human X chromosome carries between 100 and 200 different genes. The Y chromosome has less than 100 genes.

Sex-linked genes are also found in humans. For example, a recessive allele located on the X chromosome determines red–green colour-blindness. More males are colour-blind than females because females require two recessive alleles to exhibit colour-blindness. Since males have only one X chromosome, they require only one recessive allele to be colour-blind. Other sex-linked traits that affect males primarily include hemophilia, hereditary near-sightedness (myopia), and night-blindness.

This explains why **recessive lethal** X-linked disorders in humans, such as infantile spinal muscular atrophy, occur more frequently in males. This could also explain why the number of females reaching the age of 10 and beyond is greater than the number of males. Males die at birth or before the age of 10 from recessive lethal X-linked disorders.

Table 1 Possible Genotypes for *Drosophila*

Females	Males
X^RX^R	X^rY
X^RX^r	X^RY
X^rX^r	

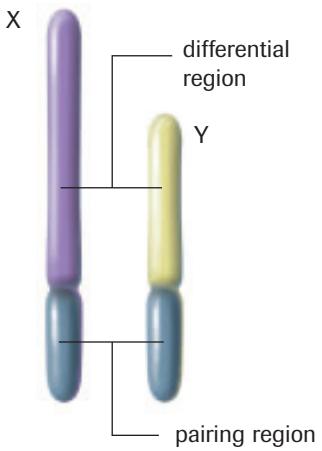

Figure 5
Sex chromosomes. Sections of the X and Y chromosomes are homologous; however, few genes are common to both chromosomes.

Barr Bodies

The difference between male and female autosomal (non-sex) cells lies within the X and Y chromosomes. Dr. Murray Barr, working at the University of Western Ontario in London, recognized a dark spot in some of the somatic cells of female mammals during the interphase of meiosis. This spot proved to be the sex chromatin, which results when one of the X chromosomes in females randomly becomes inactive in each cell. This dark spot is now called a **Barr body** in honour of its discoverer. This discovery revealed that not all female cells are identical; some cells have one X chromosome inactive, while some have the other. This means that some cells may express a certain trait while others express its alternate form, even though all cells are genetically identical. For example, if a human female is heterozygous for the skin disorder *anhidrotic ectodermal dysplasia*, she will have patches of skin that contain sweat glands and patches that do not. This mosaic of expression is typical of X chromosome activation and inactivation. In normal skin, the X chromosome with the recessive allele is inactivated and sweat glands are produced. In the afflicted skin patches, the X chromosome with the recessive allele is activated and no sweat glands are produced.

recessive lethal a trait that, when both recessive alleles are present, results in death or severe malformation of the offspring. Usually, recessive traits occur more frequently in males.

Barr body a small, dark spot of chromatin located in the nucleus of a female mammalian cell

EXTENSION

Barr Body Formation

Listen to a discussion of the formation of Barr bodies and mosaic phenotypes in females.

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LAB EXERCISE 19.A

Tracing the Hemophilia Gene

Report Checklist

- ○ Purpose
- ○ Problem
- ○ Hypothesis
- ○ Prediction
- ○ Design
- ○ Materials
- ○ Procedure
- ○ Evidence
- ● Analysis
- ○ Evaluation
- ○ Synthesis

A pedigree chart provides a means of tracing the inheritance of a particular trait from parents through successive generations of offspring. Hemophilia A is a blood-clotting disorder that occurs in about one in 7000 males. The disorder is associated with a recessive gene located on the X chromosome, normally represented as X^h. Normal blood clotting is controlled by a dominant gene, X^H. The fact that a female must inherit one of the mutated alleles from her mother and another of the mutated alleles from her father helps explain why this disorder is very rare in females. Males, on the other hand, only need to inherit one recessive allele to express the disorder.

Purpose

To use pedigree charts to trace the hemophilia gene from Queen Victoria

Evidence

See **Figure 6**.

Analysis

1. Study the pedigree chart of Queen Victoria and Prince Albert (**Figure 6**). Note the legend at top right.

(a) Who was Queen Victoria's father?

(b) How many children did Queen Victoria and Prince Albert have?

2. Locate Alice of Hesse and Leopold, Duke of Albany, on the pedigree chart.

(c) Using the legend, provide the genotypes of both Alice of Hesse and Leopold.

3. Locate the royal family of Russia on the pedigree chart by finding Alexandra. Alexandra, a descendant of Queen Victoria, married Nikolas II, Czar of Russia. Nikolas and Alexandra had four girls (only Anastasia is labelled), and one son, Alexis.

(d) Explain why Alexis was the only child with hemophilia.

(e) Is it possible for a female to be hemophilic? If not, explain why not. If so, identify a male and female from the pedigree chart who would be capable of producing a hemophilic, female offspring.

(f) On the basis of probability, calculate the number of Victoria's and Albert's children who would be carriers of the hemophilic trait.

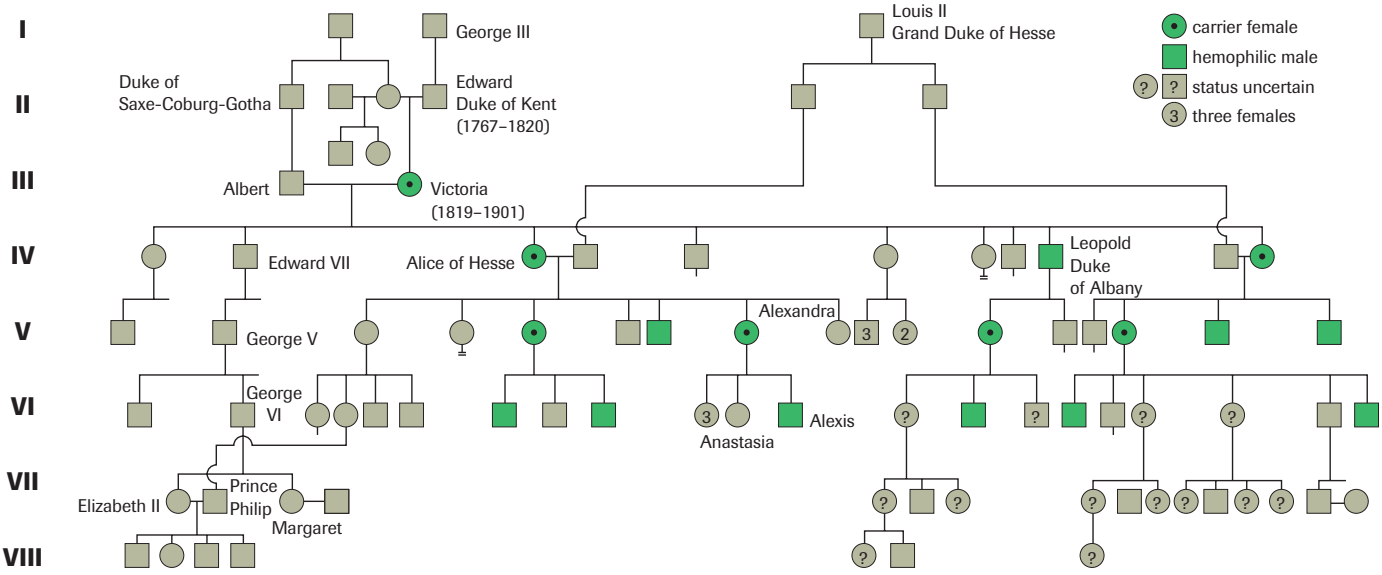

Figure 6

EXPLORE an issue

Report Checklist

- Issue
- Design
- Analysis
- Resolution
- Evidence
- Evaluation

Screening for Genetic Disorders

Screening for inherited diseases can be carried out by various methods, including detailed pedigrees and biochemical testing for known disorders. Prenatal ("before birth") diagnosis can determine the presence of many genetic conditions in the unborn fetus. Amniocentesis involves the extraction of a small sample of fluid from the amnion, the membranous sac around the fetus. Chorionic villi sampling (CVS) involves withdrawing cells from the chorion, a fluid-filled membranous sac that surrounds the amnion. CVS can yield results earlier than amniocentesis, as early as in the ninth week of pregnancy.

Before the development of a process that permitted the extraction of insulin from animals, the children of parents who passed on two copies of the recessive allele for diabetes died at a young age. Today, genetic screening can tell potential parents if they carry this allele (**Figure 7**). Huntington disease is a neurological disorder caused by a dominant allele that only begins to express itself later in life. The disease is characterized by the rapid deterioration of nerve control, eventually leading to death. Early detection of this disease by genetic screening is possible.

By having knowledge of a genetic disorder prior to birth, parents will have the opportunity to be better prepared to cope with any additional challenges the disorder may bring. Some parents may choose to terminate a pregnancy based on the results of genetic screening. This use of genetic screening is controversial.

- In small groups, research the issue of using genetic screening to detect inherited conditions. Find other ways of dealing with genetic disorders instead of genetic screening. You may wish to focus your research on one of the conditions described above.
- List the points and counterpoints against genetic screening uncovered by your group. After considering each of these, and any alternative means of dealing with genetic disorders that you found, write a statement that outlines your group's position on this issue.
- Prepare to defend your group's position in a class discussion.

Figure 7
A genetic counsellor helps a couple to assess their risks of having children with inherited diseases.

EXTENSION

The Pros and Cons of Genetic Screening

This audio clip discusses some of the advantages and disadvantages associated with genetic screening practices in humans.

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WWW WEB Activity

Simulation—Amniocentesis

Amniocentesis involves removing cells from the amniotic fluid, without damaging the fetus. Watch this animated simulation of amniocentesis to see how the cells are gathered and how they are used.

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INVESTIGATION 19.1 *Introduction*

Sex-Linked Traits

In this activity, you will cross *Drosophila* that carry genes for sex-linked traits, using virtual fruit fly software. To determine if a trait is sex-linked, you will perform two sets of crosses. In the first set of crosses, you will confirm that a trait is sex-linked using males and females with and without a trait. How will you set up the crosses to get the data you will need? In the second set of crosses, you will determine the phenotypic ratios in offspring of the F_1 generation and observe the frequency of one trait in the male and in the female offspring. What ratio would you expect for a sex-linked trait?

Report Checklist

● Purpose	● Design	● Analysis
○ Problem	○ Materials	● Evaluation
● Hypothesis	○ Procedure	○ Synthesis
● Prediction	● Evidence	

To perform this investigation, turn to page 652.

SUMMARY *Chromosomes and Genetics*

- The chromosomal theory of inheritance:
  - Chromosomes carry genes, the units of heredity.
  - Each chromosome contains many different genes.
  - Paired chromosomes segregate during meiosis. Each sex cell or gamete has half the number of chromosomes found in a somatic cell.
  - Chromosomes assort independently during meiosis. This means that each gamete receives one member from each pair of chromosomes, and that each chromosome pair has no influence on the movement of any other chromosome pair.
- Females have two X chromosomes. Males have one X and one Y chromosome.
- Sex-linked traits are controlled by genes located on the sex chromosomes. A recessive trait located on the X chromosome is more likely to express itself in males than in females, since males need only one copy of the recessive allele while females need two.
- Female somatic cells can be identified by Barr bodies, which are actually dormant X chromosomes.

Section 19.1 Questions

1. Describe how the work of Walter S. Sutton and Theodor Boveri advanced our understanding of genetics.
2. How do sex cells differ from somatic cells?
3. Describe how Thomas Morgan's work with *Drosophila* advanced the study of genetics.
4. Identify two different sex-linked traits in humans.
5. What are Barr bodies?
6. A recessive sex-linked allele *(h)* located on the X chromosome increases blood-clotting time, causing hemophilia.
   (a) With the aid of a Punnett square, explain how a hemophilic offspring can be born to two normal parents.
   (b) Can any of the female offspring develop hemophilia? Explain.
7. In humans, the recessive allele that causes a form of red–green colour-blindness *(c)* is found on the X chromosome.
   (a) Identify the F_1 generation from a colour-blind father and a mother who is homozygous for colour vision.
   (b) Identify the F_1 generation from a father who has colour vision and a mother who is heterozygous for colour vision.
   (c) Use a Punnett square to identify parents that could produce a daughter who is colour-blind.

Gene Linkage and Crossover 19.2

It is often said that great science occurs when good questions are asked. Like Mendel, Morgan asked great questions when he observed a few unexpected gene combinations when he performed some dihybrid crosses with *Drosophila*. Morgan had found a number of obvious mutations in *Drosophila*. He had noted a number of genes in *Drosophila* that had different alleles that were easy to observe, which he used in many genetic experiments. When he carried out dihybrid crosses of *Drosophila*, Morgan observed that in some of the crosses, almost all the offspring had the same combination of traits as did the parents. Morgan's hypothesis to explain these observations, which he tested with further experiments, gave further support to the theory that the genes are located on chromosomes.

Morgan first crossed *Drosophila* homozygous for wild-type body-colour (*AA*) and straight wings (*BB*) with *Drosophila* homozygous for black body-colour (*aa*) and curved wings (*bb*). The resulting F_1 generation was therefore heterozygous for both traits (*AaBb*). When members of the F_1 generation mated among themselves, the F_2 generation showed far less variability than expected. Since this was a dihybrid cross, Morgan had predicted that the F_2 generation would have a 9:3:3:1 phenotypic ratio, as was observed in the work of Mendel. Instead, nearly all the individuals with wild-type body-colour had straight wings and nearly all those with black body-colour had curved wings.

Why did the observed ratios differ so much from the predicted ratio? From these observations, Morgan concluded that the two genes must not have undergone independent segregation. For this to be true, both genes would have to be located on the same chromosome. In other words, the genes for body colour and wing shape must be linked genes.

Figure 1 illustrates what would happen to the alleles in this cross during meiosis, if Morgan's hypothesis was correct and the genes for body colour and wing shape were linked genes.

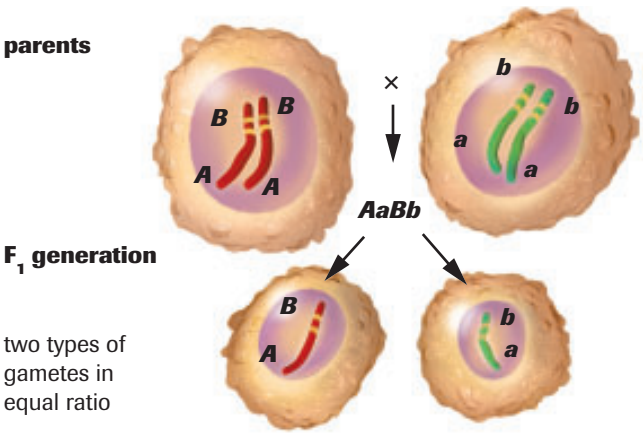


Figure 1
During meiosis, homologous chromosomes (represented as green and red chromosomes) move to opposite poles. One gamete carries the *AB* alleles and the other carries the *ab* alleles.

When two gametes from this cross unite, the new individual is heterozygous for both traits (*AaBb*). Remember that one parent carried the dominant alleles of the two linked genes (*A* is linked to *B*) and the other parent carried the recessive alleles (*a* is linked to *b*). Morgan, therefore, predicted that the F_2 generation would have a 3:1 phenotypic ratio (three flies with wild-type body-colour and straight wings to every one with black body-colour and curved wings), as shown in **Figure 2**, on the next page.

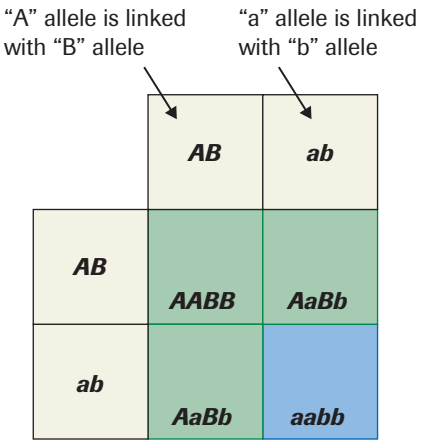


Figure 2
Punnett square analysis, assuming that all the gametes carry the same alleles as the parent. The expected phenotypic ratio is three wild-type body-colour, straight wings to one black body-colour, curved wings.

Morgan was able to find a number of linked genes. Some of these are shown in **Table 1**.

Table 1 Linked Genes Identified by Morgan's Research on *Drosophila*

Trait	Dominant/Recessive	Location
wingless *(wg)*	recessive lethal (all wingless offspring are born dead)	chromosome 2
curly wings *(Cy)*	dominant	chromosome 2
purple eyes *(pr)*	recessive nonlethal	chromosome 2
stubble bristles *(Sb)*	dominant	chromosome 3
ebony body *(e)*	recessive nonlethal	chromosome 3
miniature wings *(m)*	sex-linked recessive	chromosome 4
cut wings *(ct)*	sex-linked recessive	chromosome 4
white eyes *(w)*	sex-linked recessive	chromosome 4
vermillion eyes *(v)*	sex-linked recessive	chromosome 4

Crossing Over

Mendel had explained most of his observations by hypothesizing that the two genes were both on the same chromosome. By the Punnett square analysis shown in **Figure 2**, only two different phenotypes are predicted for these linked genes. This was not what Morgan observed. In a small number of flies from the dihybrid cross, the offspring had a different combination of traits than the parents. **Table 2** shows the numbers of the different phenotypes and their predicted genotypes. Where did the new allele combinations come from? Where did the new combinations of the two traits come from?

Table 2 Observed Progeny (F_2) of *AaBb* × *AaBb* F_1 Parents

Phenotype	Number	Possible genotype
wild-type body-colour, straight wings	290	*AABB* or *AaBb*
black body-colour, curved wings	92	*Aabb*
wild-type body-colour, curved wings	9	*AAbb* or *Aabb* indicated recombinations
black body-colour, straight wings	9	*AaBB* or *aaBb* indicated recombinations

Recall that chromosomes sometimes undergo crossing over during meiosis. During crossing over, a segment of DNA on one homologous chromosome is exchanged with the corresponding segment on the other homologous chromosome (**Figure 3**), recombining the set of genes on the chromosomes. Crossing over occurs in meiosis, during synapsis. Through crossing over, the gene combinations on a single chromosome can be altered as it is passed from generation to generation. In this cross, gametes with the gene combination *Ab* and *aB* would not occur without crossing over.

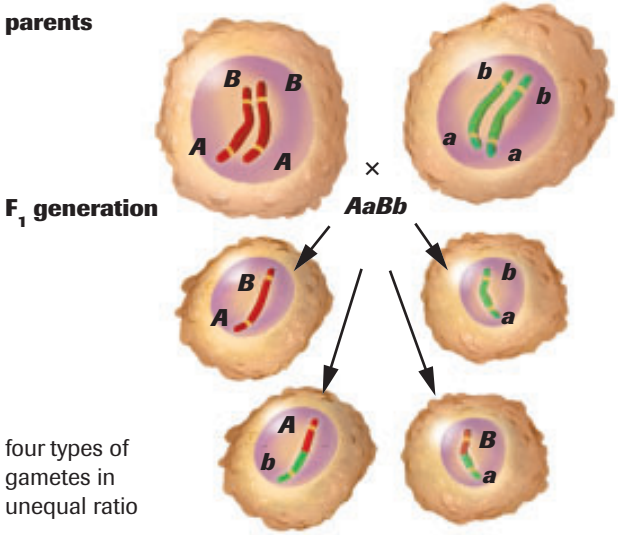


Figure 3
Consider the green chromosome to have been inherited from the father and the red from the mother. In the gametes, a chromosome that has undergone crossing has sections that are maternal (coming from the mother) and sections that are paternal (coming from the father). When the maternal and paternal homologous chromosomes carry different alleles, they may exchange alleles.

Mapping Chromosomes

As other traits in *Drosophila* were studied, it became clear that there were groups of linked genes. These **linkage groups** corresponded to different chromosomes. Furthermore, particular genes were always found at the same location (**locus**) on the chromosome. If this were not true, crossing over would not result in the exact exchange of alleles.

linkage group a group of linked genes on a chromosome

locus (plural, **loci**) a specific location along a chromosome where a particular gene is found

Morgan's experiments also showed that the frequency of crossovers between any two genes in a linkage group was always the same. The frequency of crossing over between any two genes can be stated as a percent:

$$\text{crossover percentage} = \frac{\text{number of recombinations}}{\text{total number of offspring}} \times 100\ \%$$

The crossover percentage in the offspring shown in **Table 2**, on the previous page, is

$$\text{crossover percentage} = \frac{18}{400} \times 100\ \%$$

$$= 4.5\ \%$$

The percentage of crossovers is related to the actual physical distance of the two genes on the chromosome. Genes located farther away from one another cross over at higher frequencies than genes located close together. Two genes with a crossover percentage of 1 % are much closer to one another than two genes with a crossover percentage of 12 %. Armed with this knowledge, geneticists were able to build a map of the chromosomes of *Drosophila* (**Figure 4**, next page).

When genes are in the correct order on a chromosome map, the map distances between the different genes is additive. This fact allows us to place genes in their proper order, based on the percentage crossover values between the different genes.

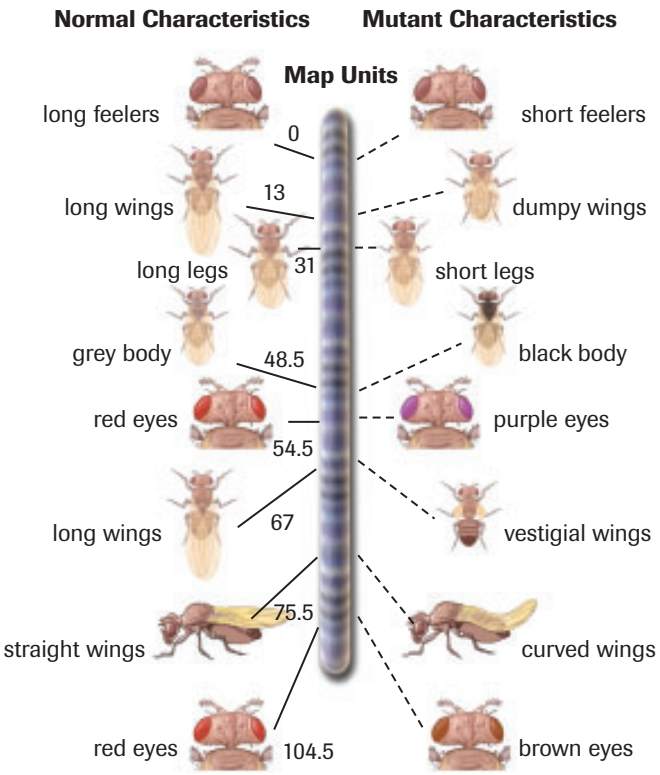


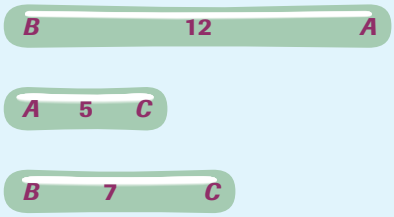
Figure 4
Gene mapping of chromosome 2 for *Drosophila melanogaster*. Note that many genes are located on one chromosome.

SAMPLE exercise 1

From crosses between different *Drosophila*, a geneticist finds that the crossover frequency between gene *A* and gene *B* is 12 %, the crossover frequency between gene *B* and gene *C* is 7 %, and between gene *A* and gene *C* is 5 %. What is the order and relative distances of these three genes on the chromosome?

Solution

If gene A were in the middle, then the sum of the distances between B and A and A and C must equal the distance between B and C. These distances are not equal, so A is not in the middle **(Figure 5)**.

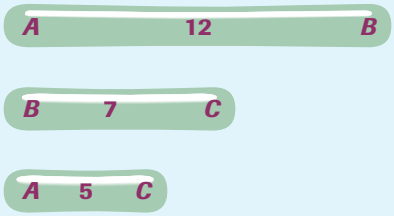


$BA + AC \neq BC$
$12 + 5 \neq 7$

Therefore, *A* is not in the middle.

Figure 5

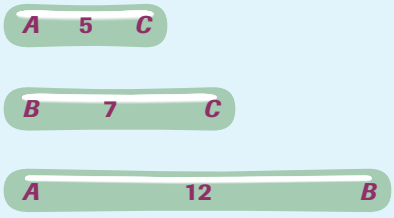
If gene B were in the middle, then the sum of the distances between A and B and between B and C must equal the distance between A and C. These distances are not equal, so B is not in the middle (**Figure 6**, next page).



$AB + BC \neq AC$
$12 + 7 \neq 5$

Therefore, *B* is not in the middle. **Figure 6**

If gene C were the middle gene, then the sum of the distances between A and C and C and B must equal the distance between A and B. These distances are equal. Therefore, C is in the middle **(Figure 7)**.



$AC + CB = AB$
$5 + 7 = 12$

Therefore, *C* is in the middle. **Figure 7**

Practice

1. A geneticist observes that the crossover frequency between gene A and gene B is 4 %, the crossover frequency between gene B and gene C is 14 %, and between gene A and gene C is 10 %. What is the order and relative distances of these three genes on the chromosome?

LAB EXERCISE 19.B

Mapping Chromosomes

A. H. Sturtevant, a student who worked with Thomas Morgan, hypothesized that

- genes are located in a linear series along a chromosome, much like beads on a string,
- genes that are closer together will be separated less frequently than those that are far apart,
- and that crossover frequencies can be used to construct gene maps.

Sturtevant's work with *Drosophila* helped establish techniques for chromosome maps.

Report Checklist

○ Purpose	○ Design	● Analysis
○ Problem	○ Materials	○ Evaluation
○ Hypothesis	○ Procedure	○ Synthesis
○ Prediction	● Evidence	

Procedure

1. Examine the picture of a chromosome (**Figure 8**, next page). Crossing over takes place when breaks occur in the chromatids of homologous chromosomes during meiosis. The chromatids break and join with the chromatids of homologous chromosomes. This causes an exchange of alleles between chromosomes.

LAB EXERCISE 19.B *continued*

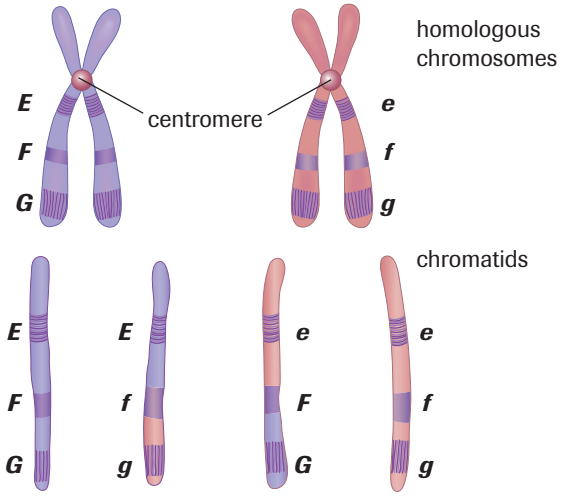


Figure 8
Crossing over

(a) Circle the areas of the chromatids that show crossing over.

(b) Using the diagram above, which genes appear farthest apart? (Choose from *EF*, *FG*, or *EG*.)

(c) Which alleles have been exchanged?

2. In 1913, Sturtevant used crossover frequencies of *Drosophila* to construct chromosome maps. To determine map distances, he arbitrarily assigned one recombination for every 100 fertilized eggs. For example, genes that had a crossover frequency of 15 % were said to be 15 units apart. Genes that had a 5 % recombination rate were much closer. These genes are 5 units apart.

(d) Using the data in **Table 3**, determine the distance between genes *E* and *F*.

Table 3

Cross	Offspring	Frequency (%)
EF × *ef*	*EF* + *ef* (from parent)	94
	Ef + *eF* (recombination)	6

(e) Would the distance between genes *e* and *f* be identical?

3. Use the data in **Table 4** to construct a complete gene map.

(f) What is the distance between genes *E* and *G*?

(g) What is the distance between genes *F* and *G*?

Table 4

Cross	Offspring	Frequency (%)
EF × *ef*	*EF* + *ef* (from parent)	94
	Ef + *eF* (recombination)	6
EG × *eg*	*EG* + *eg* (from parent)	90
	Eg + *eG* (recombination)	10
FG × *fg*	*FG* + *fg* (from parent)	96
	Fg + *fG* (recombination)	4

Analysis

(h) What mathematical evidence indicates that gene *F* must be found between genes *E* and *G*?

(i) Draw the gene map to scale. (Use 1 cm to represent 1 unit.)

(j) For a series of breeding experiments, a linkage group composed of genes *W*, *X*, *Y*, and *Z* was found to show the gene combinations in **Table 5**. (All recombinations are expressed per 100 fertilized eggs.)

Table 5

Genes	*W*	*X*	*Y*	*Z*
W	-	5	7	8
X	5	-	2	3
Y	7	2	-	1
Z	8	3	1	-

Construct a gene map. Show the relative positions of each of the genes along the chromosome and indicate distances in map units.

(k) For a series of breeding experiments, a linkage group composed of genes *A*, *B*, *C*, and *D* was found to show the gene combinations in **Table 6**. (All recombinations are expressed per 100 fertilized eggs.) Construct a gene map. Show the relative positions of each of the genes along the chromosome and indicate distances in map units.

Table 6

Genes	*A*	*B*	*C*	*D*
A	-	12	15	4
B	12	-	3	8
C	15	3	-	11
D	4	8	11	-

Using Marker Genes

Earlier in the chapter, you learned that genes located on the same chromosome are usually inherited together. **Marker genes** can be used to follow the inheritance of a linked trait. Marker genes give rise to an easily identifiable phenotype and are used to trace the inheritance of other genes that are difficult to identify. The marker gene must be located on the same chromosome and, ideally, at a very small distance from the gene being traced.

marker gene a gene that confers an easily identifiable phenotype and is used to trace the inheritance of other genes that are difficult to identify; it must be located on the same chromosome, and ideally, at a very small distance from the gene being followed

Dr. Ram Mehta, president of PBR Laboratories in Edmonton, uses gene markers to identify possible gene mutations in yeast. The yeast cells are treated with agents that might alter the genetic structure of the yeast, such as various chemicals, or environmental agents such as radiation. Since the chemical structure of DNA in human chromosomes and yeast chromosomes is the same, the yeast provides a model that helps scientists to predict how any given agent may affect human chromosomes.

Normally, yeast colonies are an off-white colour. This colour is determined by a dominant gene. Pink or red colonies indicate that a mutation in this normal, dominant gene has taken place (**Figure 9**). The red and pink colour is determined by one of two marker genes that are located along different sections of the chromosome. The marker genes are expressed only when the normal, dominant gene for colour has been inactivated by a mutation. Colonies will show both pink and red colour only when crossing over has occurred. Crossing over indicates that the agent being tested broke apart the yeast chromosome containing the marker genes. Mutation rates can be calculated from the frequency with which pink or red colonies appear.

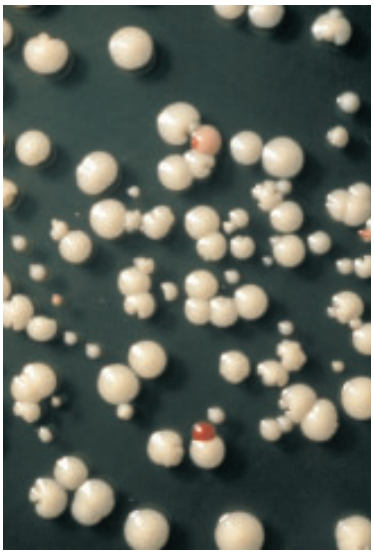


Figure 9
Mutated yeast colonies

SUMMARY — Gene Linkage and Crossover

- Linked genes do not segregate independently because they are situated on the same chromosome. Linked genes can undergo recombination due to crossing over.
- Crossing over occurs more frequently between genes located relatively far apart than for those located relatively close together.
- Genetic linkage maps can be created by sorting genes according to the percentage crossover values.

Section 19.2 Questions

1. Why does gene linkage limit the variability of an organism?
2. Does crossing over increase or decrease the variability of an organism? Explain.
3. Create a chromosome map for each set of three genes from the given information.
   (a) The crossover frequency between gene A and gene B is 23 %, the crossover frequency between gene B and gene C is 11 %, and between gene A and gene C is 12 %.
   (b) The crossover frequency between gene X and gene Z is 8.5 %, the crossover frequency between gene Y and gene Z is 2.25 %, and between gene Y and gene X is 6.25 %.

19.3 DNA Is the Hereditary Material

Figure 1
DNA contains the information that ensures that pea plants produce seeds that grow into other pea plants.

continuity of life a succession of offspring that share structural similarities with those of their parents

The nucleus of every cell in your body contains deoxyribonucleic acid, or DNA. DNA is found in the cells of all organisms, from mushrooms to trees, from sponges to mammals. Scientists' fascination with DNA arises from the fact that it is the only molecule known that is capable of replicating itself. Sugar molecules, protein molecules, and fat molecules cannot build duplicates of themselves. DNA can duplicate itself, thereby permitting cell division.

Sometimes referred to as the language of life, the genetic code is contained in 46 separate chromosomes in your body. Characteristics such as your hair colour, skin colour, and nose length are all coded within the chemical messages of DNA. Packed within the DNA are all the instructions that make you unique. Unless you are an identical twin, your DNA code is distinctively one of a kind.

DNA contains instructions that ensure **continuity of life**, which we observe as similar structural traits between members of different generations. Pea plants produce seeds that grow into other pea plants because the DNA holds the chemical messages for the roots, stems, leaves, and seed pods of a pea (**Figure 1**). In a similar way, guinea pigs give birth to other guinea pigs, and humans procreate with other humans. However, you have learned that not all offspring are identical to their parents. The uniqueness of descendants can be explained by new combinations of genes and by mutations. In order to understand how genes affect the expression of an organism's traits, you will have to learn how DNA regulates the production of protein. Proteins are the structural components of cells. DNA, therefore, not only provides continuity of life, but also accounts for the diversity of life forms.

Finding the Material of Heredity

In 1869, twenty-five-year-old Swiss biochemist Friedrich Miescher extracted a viscous white substance from white blood cells deposited on the bandages of wounded soldiers. He named this slightly acidic, phosphorus and nitrogen-rich material nuclein because he found it within the nuclei of these cells. With further work, Miescher found that nuclein was comprised of both an acidic portion, which he called nucleic acid, and an alkaline portion. The alkaline portion was later determined to be protein. Several decades later, Miescher's single nucleic acid was shown to actually be two nucleic acids, one of which was renamed ribonucleic acid (RNA) and the other, deoxyribonucleic acid (DNA). Ongoing research gradually revealed the structure, function, and importance of the remarkable and complex DNA molecule and showed it to be the source of hereditary information. This knowledge in turn triggered revolutions in the biological sciences.

Early work aimed at finding the material of heredity focused on proteins as the most probable source. In 1943, Danish biologist Joachim Hammerling demonstrated that the nucleus was likely to be the region in which the hereditary material of the cell would be found. He was able to do this as a result of research involving the large single-celled green alga *Acetabularia*. This organism grows to an average length of 5 cm and has three distinct regions known as the cap, the stalk, and the foot.

Hammerling's experiments first involved cutting the cap off of some cells and the foot, which contains the nucleus, off of others. The cells whose caps were removed were able to regenerate new caps, but the cells whose feet had been removed were not able to regenerate new ones (**Figure 2**, next page). As a result, Hammerling hypothesized that the hereditary information was contained in the foot and, more specifically, the

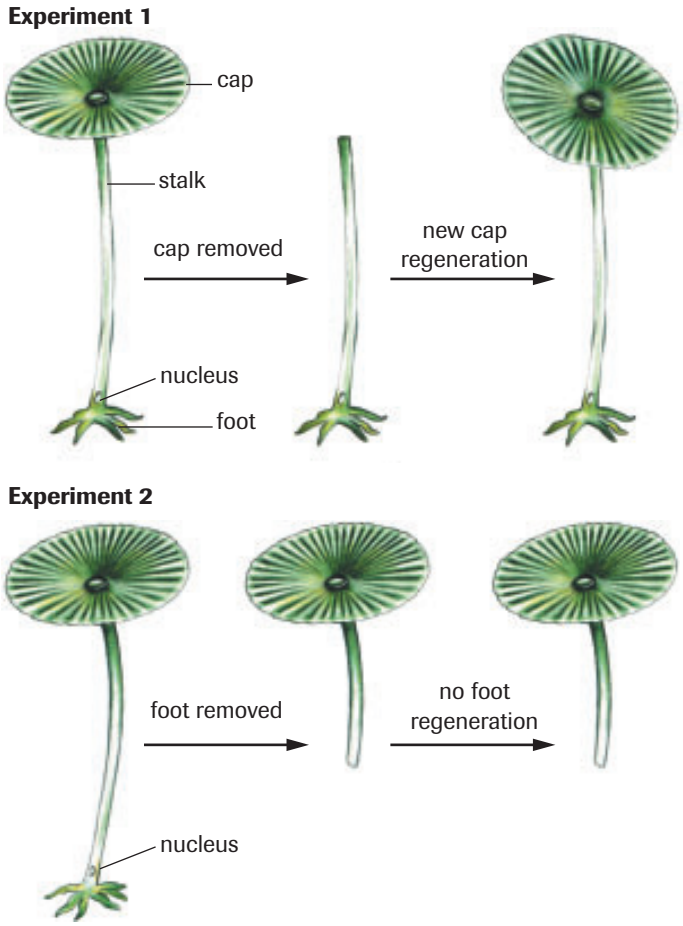


Figure 2
Hammerling's experiment strongly suggested that the hereditary material is located in the nucleus.

nucleus. To further test his hypothesis, he conducted additional experiments in which he transplanted stalks from a species of *Acetabularia* with a flowerlike cap onto the foot of another species with a disk-shaped cap. The caps that eventually developed on the transplanted stalks were all disk-shaped. Hammerling concluded that the instructions needed to build these new caps were very likely in the nucleus in the foot of the cell and not elsewhere.

Hammerling's results encouraged scientists to concentrate their search for the material of hereditary material on the nucleus and its contents. Proteins and DNA are present in the nucleus in large quantities, but DNA was initially thought to be too simple a material to account for the great variety seen in cells and cell processes, while proteins were already known to play a significant role in metabolic functions. However, work by British biologist Frederick Griffith on *Streptococcus pneumoniae*, in 1928, laid the foundation for later research. Canadian-born scientists Oswald Avery and Colin MacLeod, along with their American teammate Maclyn McCarty, built upon this work over a 14-year period culminating in 1944, and came to the conclusion that DNA was indeed the molecular material of heredity.

DID YOU KNOW?

One Man's Castle Is Another Man's Lab

Friedrich Miescher's discovery took place in the vaults of an old castle that had been converted to a laboratory. You can hear Miescher describe the process he used to isolate nuclein in an animation found by accessing the Nelson Web site.

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DID YOU KNOW?

DNA's Homes

DNA does not just reside in the nucleus. A small amount of DNA is also found in chloroplasts and mitochondria. The size of the genome varies depending on the species. Plants tend to have a larger mitochondrial genome compared with that of animals.

LAB EXERCISE 19.C

Evidence of Hereditary Material

In the 1920s, Frederick Griffith, an English medical officer, started experimenting with *Streptococcus pneumoniae*. This bacterium, which causes pneumonia, exists in two forms. One form is surrounded by a polysaccharide coating called a capsule and is known as the S form because it forms smooth colonies on a culture dish. The second harmless form has no coating and is known as the R form because it forms rough colonies on a culture dish (**Figure 3**).

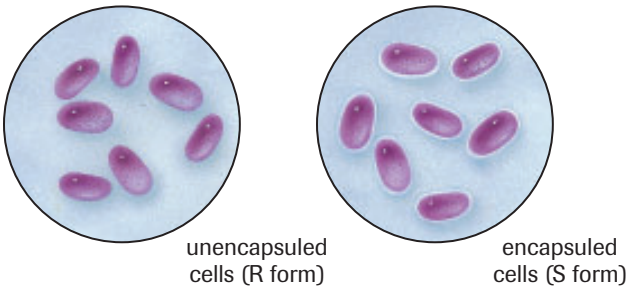

Figure 3
A representation of the two forms of *S. pneumoniae*

Report Checklist

○ Purpose	○ Design	● Analysis
○ Problem	○ Materials	● Evaluation
○ Hypothesis	○ Procedure	● Synthesis
○ Prediction	○ Evidence	

The following is an abbreviated summary of Griffith's procedures and results:

Procedure

1. Mouse A was injected with encapsulated cells (S form), while mouse B was injected with unencapsulated cells (R form).
2. Encapsulated (S-form) pneumococcal cells were heated, killed, and then injected into mouse C (**Figure 4**).
3. The heated encapsulated (S-form) cells were mixed with unencapsulated (R-form) cells. The mixture was grown on a special growth medium. Cells from the culture medium were injected into mouse D (**Figure 4**).

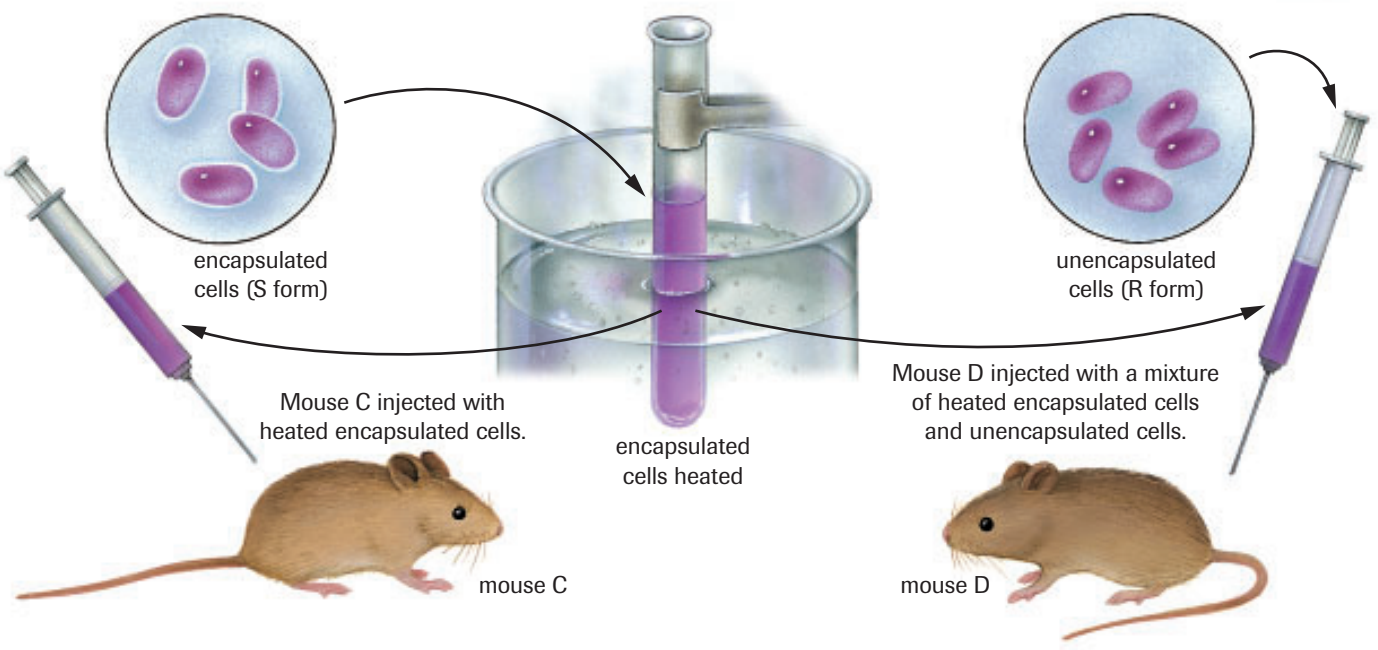

Figure 4
A visual outline of the procedure

LAB EXERCISE 19.C *continued*

Evidence

- Mouse A contracted pneumonia and died, while mouse B continued to live. Mouse B was sacrificed, and an autopsy was conducted on both mice. The autopsies revealed living S cells in mouse A's tissues and living R cells in mouse B's tissues.
- Mouse C continued to live. Mouse C was sacrificed and the autopsy revealed that no living S cells were found in the animal's tissues.
- Mouse D died. An autopsy indicated that the mouse had died of pneumonia; encapsulated (S-form) bacteria and unencapsulated (R-form) bacteria were isolated from the mouse.

Analysis and Evaluation

(a) What conclusions can you derive from the experimental results with mouse A and mouse B?

(b) Why might a scientist decide to repeat step 1 of this experimental procedure on other mice?

(c) What is the significance of the result with mouse C?

(d) Predict what would have happened to the mouse if the unencapsulated (R-form) cells had been heated and then injected. What would this step have represented in the experimental protocol?

(e) Would you have predicted that mouse D would die? Explain why or why not.

(f) A microscopic examination of the dead and live cell mixture (step 3) revealed cells with and without capsules. What influence did the heat-destroyed cells have on the unencapsulated cells?

(g) Griffith hypothesized that a chemical in the dead, heat-treated, encapsulated cells (step 3) must have altered the living unencapsulated cells and he dubbed this chemical phenomenon *transformation*. In 1944, Oswald Avery, Maclyn McCarty, and Colin MacLeod conducted experiments in test tubes with *Streptococcus pneumoniae* that led them to conclude that DNA is the *transforming principle*, as they called it, and not proteins, as was widely believed. In their experiments, what must have happened to the DNA when the cells divided?

Synthesis

(h) To discover the identity of the transforming principle, Avery and his associates ruptured heat-killed, encapsulated cells to release their contents. RNA, DNA, protein, and purified polysaccharide coats were isolated and were tested for transforming activity. Avery and his associates found that only R cells mixed with purified DNA isolated from dead S cells were transformed to S cells. When R cells were mixed with purified RNA, with the polysaccharide coat, or with protein extracted from dead S cells, only R cell colonies were isolated. Do these results support their hypothesis? Explain.

(i) Predict the experimental results of the following protocols. Support your prediction with a hypotheses.

- Polysaccharide-digesting enzymes are used to digest the encapsulated polysaccharide coat of the heated S form of the bacteria. The treated bacteria are then placed with unencapsulated pneumonia cells, which are then injected into a mouse.
- Heated encapsulated bacteria are treated with DNAase, a DNA-digesting enzyme. The treated bacteria are then mixed with unencapsulated pneumonia cells, which are injected into a mouse.
- All proteins are extracted from the heated encapsulated bacteria. The treated bacteria are then mixed with unencapsulated pneumonia cells, which are injected into a mouse.

(j) Based on the information provided, suggest improvements to the experimental protocols.

Figure 5
Dr. Oswald Avery

Figure 6
Dr. Colin MacLeod

bacteriophage a virus that infects bacteria

WEB Activity

Canadian Achievers—Avery and MacLeod

Canadian-born scientists Dr. Oswald Avery and Dr. Colin MacLeod spent their early years as scientists in Nova Scotia, where they were born. They met in New York, where, together with American scientist Maclyn McCarty, they painstakingly isolated components of pneumococci (*Streptococcus pneumoniae*) for over a decade before identifying DNA as the transforming principle. You can find more information on this classic experiment in an animation by accessing the Nelson science Web site.

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Confirming the Chemical of Heredity

Frederick Griffith's work in the 1920s began because he was trying to develop a vaccine against pneumonia caused by *Streptococcus pneumoniae*. However, his unexpected experimental observations, followed by the work of Avery, McCarty, and MacLeod, led scientists to begin questioning the initial assumption within the scientific community that the material of heredity was protein. What was now needed was experimental evidence that would clearly and conclusively indicate that DNA was indeed the material of heredity. This evidence was to come some six years after the work of Avery's team as the result of an innovative experiment.

Alfred D. Hershey and Martha Chase

It was not until 1952 that DNA was accepted as the hereditary material. That year, American scientists Alfred Hershey and Martha Chase conducted experiments using a virus (**bacteriophage** T2) that infects a bacterial host (**Figure 7**). Bacteriophages (commonly called phages) consist of two components: DNA and a protein coat. A bacteriophage infects a bacterial cell by attaching to the outer surface of the cell and injecting its hereditary information into it. This leads to the production of thousands of new viruses, which then burst out of the cell, resulting in its death. The results of Hershey and Chase's experiments showed that only the DNA from the bacteriophage, and not the protein coat, enters the bacteria to direct the synthesis of new viral DNA and new viral protein coats.

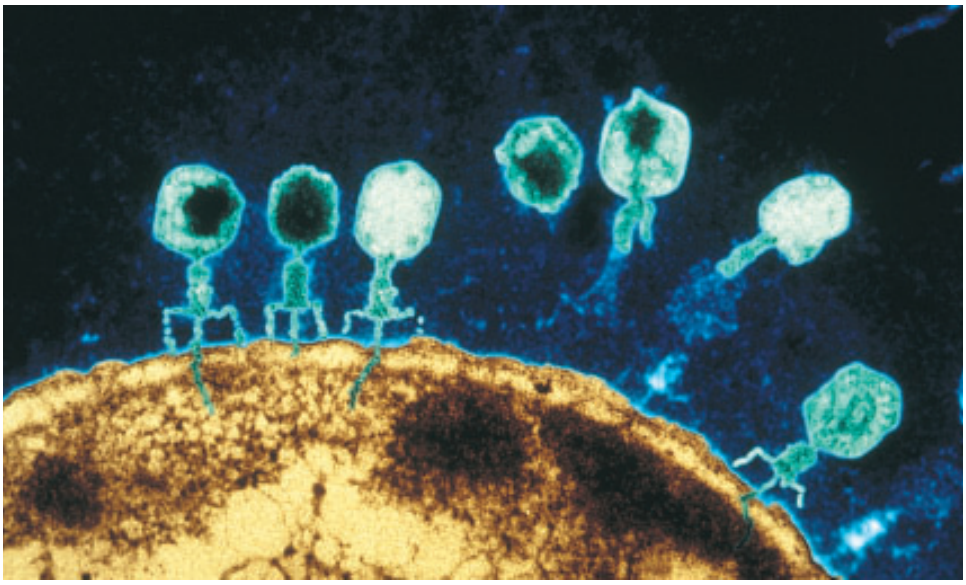

Figure 7
Micrograph of a bacteriophage injecting its DNA into a bacterial cell

Proteins contain sulfur but no phosphorus, whereas DNA contains phosphorus but no sulfur. Therefore, to track the location of DNA and proteins, Hershey and Chase tagged the viral proteins with an **isotope** of sulfur, ^{35}S, and the viral DNA with an isotope of phosphorus, ^{32}P. ^{35}S and ^{32}P are **radioisotopes** of sulfur and phosphorus, respectively. They are easy to track in an experiment because radioisotopes are unstable and the radiation that they emit as they decay can be measured.

isotope one of two or more atoms of the same element containing the same number of protons but a different number of neutrons

radioisotope an unstable isotope that decays spontaneously by emitting radiation

Each type of tagged bacteriophage was allowed to infect a separate batch of bacterial host cells and to multiply. The bacterial cells were put into a blender to remove the protein coats of the viruses from the surfaces of the bacteria. The mixtures were then subjected to centrifugation to isolate the individual components (bacteria as a pellet and viral particles in the liquid). The bacterial cells that were exposed to viruses containing radioactively labelled DNA contained ^{32}P. The bacterial cells that were exposed to viruses whose protein coats were radioactively tagged with ^{35}S did not contain any radioactivity; instead, the radioactive ^{35}S was found in the culture medium (**Figure 8**). These experiments illustrate that phosphorus-rich DNA was injected into the bacterial cells. In addition, Hershey and Chase found that the bacteriophages in both experiments reproduced and destroyed the bacterial cells that they had infected. This observation further supported the claim that DNA entering the host bacterial cell carries all the genetic information. Hershey and Chase's experiments ended the debate. DNA was accepted as the hereditary material.

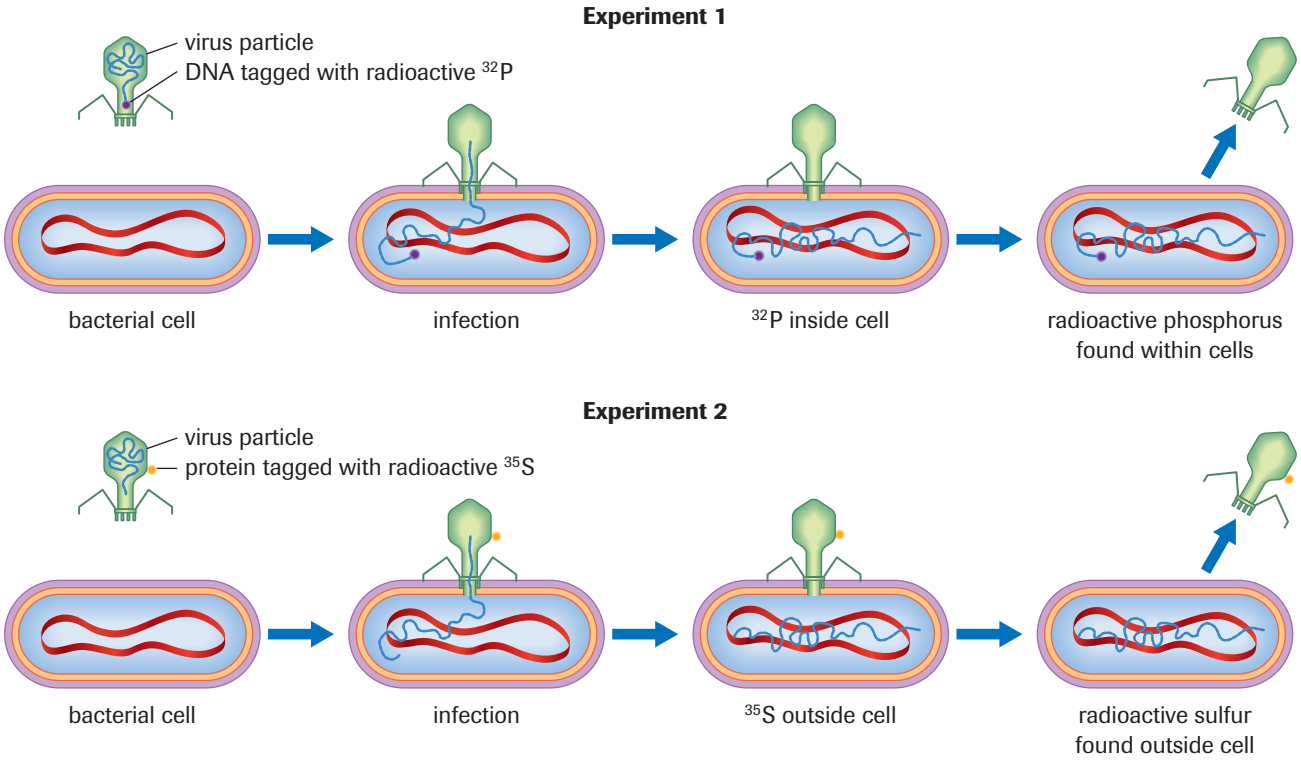

Figure 8
Hershey and Chase's experiment conclusively showed that DNA was the hereditary material.

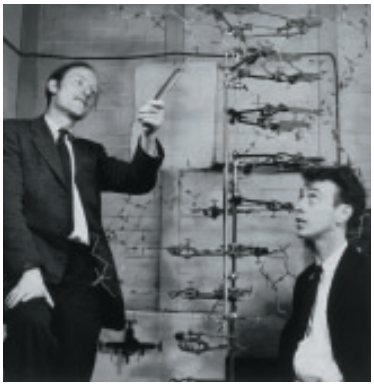

Figure 9
Francis Crick and James Watson were awarded the Nobel Prize for Physiology or Medicine in 1962 for deducing the structure of DNA.

The Race to Reveal the Structure

When scientists confirmed that DNA was the material of heredity, their focus shifted to understanding how it works. Part of that understanding would come from knowing its structure since, as in other subjects, structure in biology provides many clues about function. In the race to be the first to discover the structure of DNA, scientists around the world employed emerging technologies to help them gain new insights into this mysterious "molecule of life." In the end, the honour would go James Watson and Francis Crick (**Figure 9**).

James Watson was considered a child prodigy when he entered the University of Chicago at the age of 15. He began studying ornithology, but eventually turned his attention to genetics and molecular biology. In 1951, he began studies at England's Cambridge University, where he met Francis Crick, a physicist who had served with the British army during World War II. Each would bring to bear his experience from a different area of science to interpret and synthesize the experimental data that were rapidly mounting.

One source of important data came from the Cambridge laboratory of Maurice Wilkins, where researcher Rosalind Franklin used a technique called X-ray diffraction to help determine the structure of the DNA molecule. Another source of data involved the comparison of the chemical structure of DNA molecules in different organisms. By this time it had long been known that DNA is comprised of chains of molecules called **nucleotides**. The nucleotides consist of a 5-carbon cyclic ring structure called a **deoxyribose sugar** (**Figure 10**) having one of four **nitrogenous bases** attached to its 1′ carbon and a **phosphate group** attached to it 5′ carbon (**Figure 11**). The carbons in the sugar are identified by the numbers one to five and a prime (′) symbol to distinguish them from the carbons in the nitrogenous base. The four nitrogenous bases are adenine (A), guanine (G), thymine (T), and cytosine (C). Adenine and guanine are double-ringed structures classed as purines, while thymine and cytosine are single-ringed structures classed as pyrimidines. The only difference in the nucleotides is in their bases.

nucleotide a molecule having a five-carbon sugar with a nitrogenous base attached to its 1′ carbon and a phosphate group attached to its 5′ carbon

deoxyribose sugar a sugar molecule containing five carbons that has lost the –OH (hydroxyl group) on its 2′ position

nitrogenous base an alkaline, cyclic molecule containing nitrogen

phosphate group a group of four oxygen atoms surrounding a central phosphorus atom found in the backbone of DNA

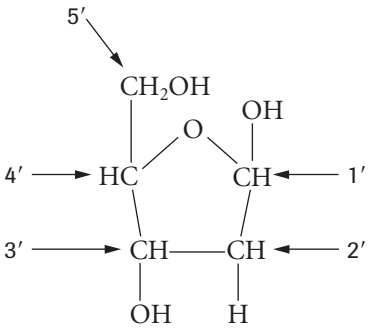

Figure 10
A deoxyribose sugar with numbered carbons

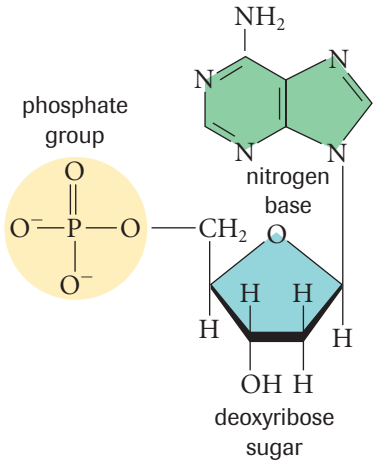

Figure 11
A DNA nucleotide is comprised of a deoxyribose sugar, a nitrogenous base, which in this case is adenine, and a phosphate group.

Biochemist Erwin Chargaff's evidence was crucial to helping Watson and Crick construct an accurate model of DNA. His observations determined that, for the DNA of any given species, the amount of adenine was always equal to the amount of thymine and the amount of guanine was always equal to the amount of cytosine. This relationship between the bases was consistent across all the species that he investigated. Although one species might have a different amount of adenine compared to another species, for example, the amount of thymine in each species was always equal to the amount of adenine.

Just as crucial was the X-ray photograph taken by Rosalind Franklin, which indicated that DNA was a helix that was likely double-stranded, that the distance between the strands was constant, and that the helix completed a full turn once every ten base pairs (**Figure 12**). Given this new data, Watson and Crick were able construct a three-dimensional scale model of DNA that portrayed the relationship between the bases as well as all of the nucleotide chemical bond angles and spacing of atoms consistent with the observations of other researchers. They presented their model to the scientific community in 1953, and in 1962 were awarded the Nobel Prize along with Maurice Wilkins. Because she had died prior to 1962 and the Nobel Prize is awarded only to living recipients, Rosalind Franklin was not included despite the acknowledgement of the significant importance of her photograph to the model proposed by Watson and Crick.

The Watson and Crick model of DNA structure is essentially the same one used by scientists today. Scientists already knew that molecules of DNA were made up of sugars (deoxyribose), phosphate, and four different nitrogen bases: adenine, guanine, cytosine, and thymine. What scientists did not know was the way in which these bases were arranged.

Learning Tip

Chargaff's Rules

The proportion of A always equals that of T (A = T).
The proportion of G always equals that of C (G = C).
A + G = T + C

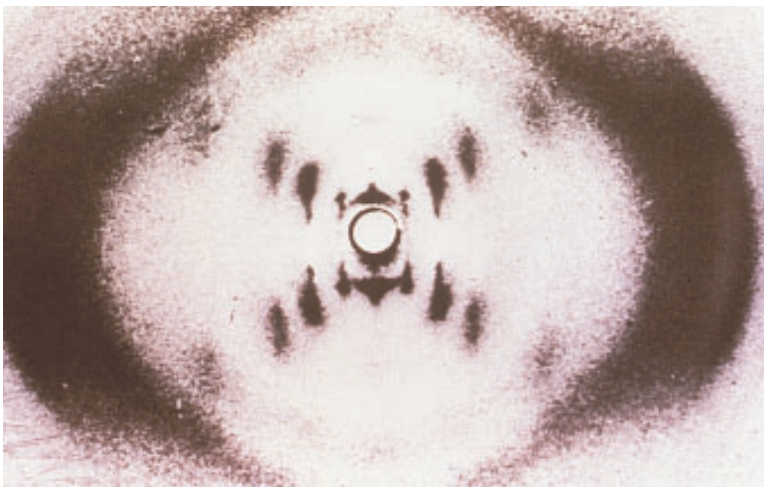

Figure 12
Rosalind Franklin's X-ray diffraction pattern of DNA revealed that it had a helical structure.

WEB Activity

Simulation—Elementary, My Dear Crick

Erwin Chargaff visited Watson and Crick in Cambridge in 1952. Crick's lack of knowledge with respect to nitrogenous bases did not impress Chargaff. By the following year, Watson and Crick had constructed their model of DNA. Enjoy Watson and Crick's deductive process in an animation found by accessing the Nelson Web site.

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Figure 13
Rosalind Franklin's X-ray crystallography was crucial to the determination of the structure of DNA.

Politics and Science

Watson and Crick might not have been credited as the co-discoverers of DNA were it not for politics. The X-ray diffraction technique developed in England had been used by Maurice Wilkins and Rosalind Franklin (**Figure 13**) to view the DNA molecule. At that time, the American scientist Linus Pauling, a leading investigator in the field, was refused a visa to England to study the X-ray photographs. Pauling, along with others, had been identified by then U.S. Senator Joseph McCarthy as a communist sympathizer for his support of the anti-nuclear movement. Many scientists today believe that the United States passport office may have unknowingly determined the winners in the race for the discovery of the double-helix model of DNA.

The McCarthy era of the early 1950s is considered by many historians as a time of paranoia and repression. Many creative people had their careers stifled or destroyed because of their perceived association with communism. In most cases the charges were unfounded. It is perhaps ironic that, in 1962, Linus Pauling was awarded a Nobel Prize, this time for his dedication to world peace.

INVESTIGATION 19.2 ***Introduction***

Isolation and Quantification of DNA

In this activity, you will extract DNA from both beef liver and onion cells in Parts 1 and 2. If your school has the necessary reagents and equipment, you will then have the option of testing for the presence of DNA in Part 3 and of determining its concentration using a spectrophotometer in Part 4. You will need to gather evidence and analyze and evaluate the results that you observe, and to then explain those results in writing. Heed all cautions and wear safety equipment as instructed.

Report Checklist

● Purpose	● Design	● Analysis
○ Problem	○ Materials	● Evaluation
○ Hypothesis	○ Procedure	○ Synthesis
○ Prediction	● Evidence	

To perform this investigation, turn to page 653.

EXPLORE an issue

Competition and Collaboration Advance Science

Scientists have been described as intelligent, ambitious, and sometimes competitive. Yet, for science to progress, many individuals must work together in a collaborative, communicative atmosphere. Current science demands two conflicting ideologies: competition and collaboration. A fine balance is not necessarily struck between the two. Other factors that come into play are economics, politics, market demand, profit, and patriotism in times of war.

Statement
Competition is the key driving force of science, followed by collaboration.

- Form groups to research this issue. Prepare a position paper in point form that supports or disputes this statement, using a specific example. Some scientists and case studies that may be used include Robert Oppenheimer's and Phillip Morrison's role in the Manhattan Project; the perception of Linus Pauling as a communist and the denial of a visa for him to visit Watson and Crick in Cambridge; Craig Venter and Eric Lander leading opposing research teams in the Human Genome Project; and Fritz Haber's role in the production of deadly gases during World War I.
- Search for information in periodicals, on CD-ROMS, and on the Internet.

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- As a group, present your supported view in a class discussion.

Issue Checklist

○ Issue	○ Design	● Analysis
● Resolution	● Evidence	● Evaluation

SUMMARY DNA is the Hereditary Material

Year	Scientist	Experimental results
late 1860s	Friedrich Miescher	• isolated nonprotein substance from nucleus of cells; named this substance nuclein
1928	Frederick Griffith	• experimented using mice and two different strains of pneumococcus bacteria (virulent and nonvirulent); observed that when heat-treated virulent pneumococcus was mixed with nonvirulent pneumococcus and was injected into healthy mice, death resulted • discovered the process of transformation
1943	Joachim Hammerling	• experimented using green alga *Acetabularia;* observed that regeneration of new appendages was driven by the nucleus-containing "foot" of the alga • hypothesized that hereditary information is stored in the nucleus
1944	Oswald Avery, Maclyn McCarty, and Colin MacLeod	• demonstrated that DNA was the transforming principle of pneumococcus bacteria
1949	Erwin Chargaff	• discovered that in the DNA of numerous organisms the amount of adenine is equal to the amount of thymine, and the amount of guanine is equal to that of cytosine
1952	Alfred Hershey and Martha Chase	• used radioactively labelled viruses, infected bacterial cells; observed that the infected bacterial cells contained radioactivity originating from DNA of the virus, suggesting that DNA is hereditary material
1953	Rosalind Franklin	• produced an X-ray diffraction pattern of DNA that suggested it was in the shape of a double helix
1953	James Watson and Francis Crick	• deduced the structure of DNA using information from the work of Chargaff, Franklin, and Maurice Wilkins

Section 19.3 Questions

1. Describe how the experiments of Joachim Hammerling; Frederick Griffith; Oswald Avery, Maclyn McCarty, and Colin MacLeod; and Alfred Hershey and Martha Chase strengthened the hypothesis that DNA is the hereditary material.
2. Explain why Hammerling's experiment cannot be used as conclusive scientific evidence that DNA is the hereditary material.
3. Hammerling chose *Acetabularia* as a model organism for his experiment. Identify some of the characteristics of this green alga that rendered it an ideal organism. Scientists use model organisms in many of their experiments. Identify social, economic, and physical characteristics that would make an organism highly suitable for experimental research. Explain why humans do not make ideal research subjects.
4. Explain why it is important to study both the historic experiments that revealed genetic principles and the principles themselves. Support your reasons, using examples.
5. It can be argued that the repetition of experiments is a waste of time, money, and other valuable resources. Provide arguments that support and dispute this statement. Use examples from the experiments of Griffith and of Avery, McCarty, and MacLeod to strengthen your arguments.

INVESTIGATION 19.1

Sex-Linked Traits

In this activity, you will cross *Drosophila* (**Figure 1**) that carry genes for sex-linked traits using virtual fruit fly software. To determine if a trait is sex-linked, you will perform two sets of crosses: A and B.

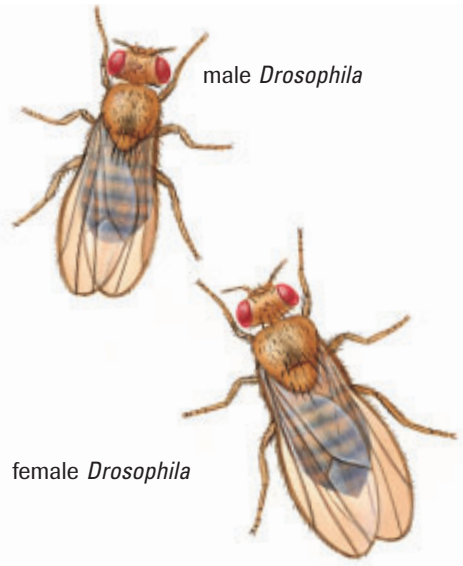

Figure 1
Drosophila males are smaller and have a rounded abdomen while the larger females have a pointed abdomen.

Familiarize yourself with the software before starting this activity. Start with the tutorial. Note that the labelling of traits in the software is different from the conventions used in this textbook. Be sure you understand what each label in the software correlates to in the textbook.

For cross A, these conditions must be met if the trait is sex-linked:

- In the F_1 generation, female offspring inherit the trait of the male parent and male offspring inherit the trait of the female parent.
- In the F_2 generation, there is a 1:1 phenotypic ratio for the traits in both males and females.

For cross B, you will confirm that the trait is sex-linked. You will cross parents with traits that are opposite to the traits of the parents in cross A. By examining the phenotypic ratios in offspring of the F_1 generation, you can observe the greater frequency of one trait in either the male or the female offspring.

Report Checklist

● Purpose	○ Design	● Analysis
○ Problem	○ Materials	● Evaluation
● Hypothesis	○ Procedure	○ Synthesis
● Prediction	● Evidence	

Problems

If white eye colour in *Drosophila* is a sex-linked recessive trait, what are the phenotypic ratios of the F_1 generation when a homozygous red-eyed female and a white-eyed male are crossed?

What other traits are sex-linked in *Drosophila*? Are they recessive or dominant?

Materials

virtual fruit fly simulation software
computers

Procedure

1. Log onto the software. Remember that each parent is homozygous for the trait chosen.
2. Select 1000 offspring.
3. For crosses A and B, follow these algorithms:
   A: P: white-eyed female × red-eyed male
   F_1: red-eyed female × red-eyed male
   F_2: red-eyed female × white-eyed male
   B: P: homozygous red-eyed female × white-eyed male
   F_1: red-eyed female × red-eyed male
4. For cross A, create a Punnett square to show the expected phenotypic ratio of offspring in each generation. Also, be sure to indicate the genotype of each phenotype.
5. After cross A, count the flies and the number of offspring (out of 1000) of each sex and with each trait. Record the information in a table beside the corresponding Punnett square.
6. When you have finished cross A, create a new parental generation.
7. Carry out cross B. Follow steps 4 to 6.
8. Determine if other traits are sex-linked. Follow the same procedure as in step 3, using new traits. Indicate which traits you are examining.

INVESTIGATION 19.1 *continued*

Analysis

(a) In one or two paragraphs, describe the results of crosses A and B. Is white eye colour in *Drosophila* sex-linked? If so, which sex does this trait appear in more frequently? Explain.

(b) In one or two paragraphs, describe the results with the other traits you examined. Is the trait sex-linked? If so, which sex does this trait appear in more frequently? Is the trait recessive or dominant? Explain.

Evaluation

(c) List and briefly explain any technical difficulties you had using the software.

(d) What improvements would you suggest to enhance the usefulness of the software?

(e) What are the advantages of using software to carry out this investigation compared to conducting it with actual *Drosophila*?

INVESTIGATION 19.2

Isolation and Quantification of DNA

Report Checklist

● Purpose	● Design	● Analysis
○ Problem	○ Materials	● Evaluation
○ Hypothesis	○ Procedure	○ Synthesis
○ Prediction	● Evidence	

In Parts 1 and 2 of this investigation, you will isolate DNA from onion cells and beef liver. Part 3 verifies the presence of DNA in your extraction using a biological analysis and Part 4 quantifies the amount of DNA using spectrophotometry. Parts 3 and 4 are optional depending on whether your school has the necessary reagents.

Problem

How much DNA can be extracted from plant and animal cells using simple laboratory methods?

Materials

safety goggles
rubber gloves
fresh beef liver
scissors
mortar and pestle
0.9 % (w/v) solution of sodium chloride (NaCl)
three 10 mL graduated cylinders
sand (very fine, washed)
cheesecloth
two 50 mL beakers, or two large test tubes
10 % (w/v) solution of sodium dodecyl sulfate (SDS)
95 % ethanol (chilled)
50 mL graduated cylinder
glass rod
four medium test tubes
4 % (w/v) solution of sodium chloride (NaCl)
onion
blender (optional)
hot-water bath
boiling chips
ice-water bath
meat tenderizer solution (3 g/50 mL of solution)
diphenylamine solution
25 mL graduated cylinder
Pasteur pipette, or plastic graduated eyedropper
distilled water
DNA standard solution
test-tube rack
spectrophotometer
cuvette
facial tissue

Procedure

DNA extraction is the first step in many biotechnological procedures. Cell walls and cell membranes must be disrupted to isolate DNA. In addition, lipids, proteins, and sugars must be separated from nucleic acid. In the following procedure, heat, detergents, salts, and cleaving enzymes are used to minimize contamination from nonnucleic acid molecules and to maximize purification.

Part 1: Extraction of DNA from Beef Liver

The ethanol solution is toxic and flammable. Keep it away from all sources of heat.

1. Obtain a 10 g to 15 g sample of beef liver and place it in the mortar.
2. Using scissors, cut the liver into small pieces.

INVESTIGATION 19.2 *continued*

3. Add 10 mL of 0.9 % NaCl solution to the diced liver. Use a 10 mL graduated cylinder to measure out the NaCl. Add a pinch of sand into the mixture to act as an abrasive, and grind the tissue thoroughly for approximately 5 min.
4. Strain the liver cell suspension through several layers of cheesecloth to eliminate any unpulverized liver. Collect the filtrate into a 50 mL beaker.
5. Add 3 mL of 10 % SDS solution. If a centrifuge is available, spin the suspension, and remove and save the supernatant. If a centrifuge is not available, mix the suspension thoroughly for 30 s and proceed to step 6.
6. Gently layer twice the volume (approximately 25 mL) of cold 95 % ethanol on the supernatant as that of the total volume of the cell suspension–SDS mixture. Use a 50 mL graduated cylinder to measure out the ethanol.
7. Using the glass rod, stir gently and slowly. A white, mucuslike substance will appear at the interface between the solutions. This substance is the DNA–nucleoprotein complex. After the complex has formed, twirl the stirring rod slowly and collect it onto the rod. Record your observations.
8. Place the isolated DNA–nucleoprotein complex into a test tube containing 3 mL of 4 % NaCl solution for later use. Use a 10 mL graduated cylinder to measure the 4 % NaCl solution. Pour the waste alcohol into the waste alcohol container designated by your teacher.

Part 2: Extraction of DNA from Onion

Onion is used because of its low starch content, which allows for a higher purity DNA extraction.

9. Repeat steps 1 to 5 using finely chopped onion. Instead of hand chopping the onion, a blender could be substituted, which gives optimum results.
10. Stir the mixture and let it sit for 15 min in a 60 °C water bath containing boiling chips. (Any longer and the DNA starts to break down.)
11. Cool the mixture in an ice-water bath for 5 min, stirring frequently.
12. Add half the volume of meat tenderizer solution as is present in your filtrate and swirl to mix.
13. Repeat steps 6 to 8.

Part 3: Testing for the Presence of DNA

The presence of DNA may be detected qualitatively with the reagent diphenylamine. Diphenylamine reacts with the purine nucleotides in DNA, producing a characteristic blue colour.

Diphenylamine solution contains glacial acetic acid. Be very careful not to spill any of the solution on yourself or on any surface. Inform your teacher immediately if any spills occur. Wear safety goggles and rubber gloves when handling this solution.

14. Stir the DNA from the onion and beef liver with their respective glass rods to resuspend them into the 4 % NaCl solution.
15. Dispense 15 mL of diphenylamine solution into a 25 mL graduated cylinder. The teacher will direct you to the stock diphenylamine solution, which will have been set up in a burette.
16. Transfer 5 mL of the solution to a 10 mL graduated cylinder with a Pasteur pipette or with a plastic graduated eyedropper.
17. Add 5 mL of diphenylamine solution to the DNA suspension obtained from the onion and from the beef liver.
18. Repeat step 16 and add 5 mL of diphenylamine solution to a test tube containing 3 mL of distilled water (the blank).
19. Repeat step 16 and add 5 mL of diphenylamine solution to a test tube containing 3 mL of DNA standard (the standard).
20. Place all of the test tubes in a boiling water bath (containing boiling chips) for 10 min and record the colour changes. Record your observations.
21. Remove the test tubes from the hot-water bath and place into a test-tube rack. Allow the tubes to cool before proceeding.

INVESTIGATION 19.2 *continued*

Part 4: Quantitative Determination of DNA Concentration Using Spectrophotometry

The principle underlying a spectrophotometric method of analysis involves the interaction of electromagnetic (EM) radiation (light) with matter. When EM radiation strikes an atom, energy in the form of light is absorbed. The remainder of the energy passes through the sample and can be detected. The more molecules that are present, the more energy will be absorbed, resulting in a higher absorbance reading. Since the relationship is direct, we can determine the concentration of an unknown by comparing it with a known. In this case, the unknown is the concentration of DNA in your samples and the known is the DNA standard.

22. Set the spectrophotometer to a wavelength of 600 nm. (See the video *Spec 20* on the Nelson Web site.)

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23. Fill a dry cuvette with the solution that consists of the distilled water and the diphenylamine. This will serve as a blank.
24. Wipe off any fingerprints from the outside of the cuvette by holding the cuvette at the very top and using a facial tissue. Place the blank into the spectrophotometer and set the absorbance to 0.00. (See the video *Spec 20* on the Nelson Web site.)

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25. Pour the blank solution back into its original test tube and place it in a test-tube rack.
26. Rinse the cuvette with a tiny amount of standard DNA solution (DNA standard and diphenylamine from step 19). Wipe off any fingerprints in the manner described in step 24.
27. Place the DNA standard solution into the spectrophotometer, then record the absorbance. (See the video *Spec 20* on the Nelson Web site.)

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28. Pour the DNA standard solution into its original test tube and save in case of error.
29. Repeat steps 26 to 28 with the beef liver extract solution and with the onion extract solution.

Analysis

(a) Propose reasons that the onion cells required heating and the liver cells did not.

(b) DNA was spooled out using a glass rod. How do you account for the "stickiness" of DNA to glass?

(c) Describe the DNA you extracted. If DNA is a rigid structure, why do the DNA strands appear flexible? What features of DNA's structure account for its stiffness? If DNA is rigid, how does it coil tightly into a small space?

(d) Comment on the purity of the DNA extracted.

(e) Compare the amount of DNA extracted from the onion versus that from the liver. Which source of DNA provided more of the molecule? Account for this observation, given your knowledge of cell structure and given differences in the procedure.

(f) What was the purpose of the standard DNA solution? What was the purpose of the blank?

(g) Did the spectrophotometric results correlate with the qualitative observations obtained from the diphenylamine test? Comment.

(h) Calculate the amount of DNA extracted from each source using your standard as a guide.

(i) The liver and onion were chopped very finely. Provide reasoning for this step. If the step was omitted, what effect would this omission have on the results?

(j) SDS is a detergent. Describe how detergents work and explain the role of SDS in the protocol.

(k) How does NaCl contribute to maximum DNA extraction? (Hint: Think about DNA's chemical constituents.) Keep in mind that NaCl is a salt that ionizes in solution.

(l) What is the purpose of adding cold ethanol to each extraction? How does this phenomenon work?

(m) In the extraction of DNA from onion, you added a meat tenderizer solution. The meat tenderizer solution contains an enzyme called papain. What role does papain play in the extraction?

(n) Identify three properties of DNA that are demonstrated by this investigation.

Evaluation

(o) Suggest possible sources of error in this procedure and describe their effect on the results.

Chapter 19 SUMMARY

Outcomes

Knowledge

- summarize the historical events that led to the discovery of the structure of the DNA molecule, as demonstrated by Franklin, Watson, and Crick (19.3)
- explain the limitations of variability due to gene linkage and the influence of crossing over on assortment of genes on the same chromosome (19.2)
- explain the relationship between variability and the number of genes controlling a trait (19.2)
- compare the pattern of inheritance produced by genes on the sex chromosomes to that of genes on autosomes, as investigated by Morgan and others (19.1)

STS

- explain that decisions regarding the application of scientific and technological development involve a variety of perspectives including social, cultural, environmental, ethical, and economic considerations (19.2, 19.3)

Skills

- ask questions and plan investigations (19.2, 19.3)
- conduct investigations and gather and record data and information (19.1, 19.2)
- analyze data and apply mathematical and conceptual models by analyzing crossover data for a single pair of chromosomes to create a chromosome map showing gene arrangement and relative distance (19.2)
- work as members of a team and apply the skills and conventions of science (all)

Key Terms

19.1

autosome
linked genes
sex-linked trait
recessive lethal
Barr body

19.2

linkage group
locus (loci)
marker gene

19.3

continuity of life
bacteriophage
isotope
radioisotope
nucleotide
deoxyribose sugar
nitrogenous base
phosphate group

MAKE a summary

1. Create a poster of a human genome that shows the principles of sex-linked genes and helps show the relationship between genes and chromosomes. Label the sketch with as many of the key terms as possible. Check other posters and use appropriate ideas to make your poster clear.
2. Revisit your answers to the Starting Points questions at the beginning of the chapter. Would you answer the questions differently now? Why?

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The following components are available on the Nelson Web site. Follow the links for *Nelson Biology Alberta 20–30*.

- an interactive Self Quiz for Chapter 19
- additional Diploma Exam-style Review Questions
- Illustrated Glossary
- additional IB-related material

There is more information on the Web site wherever you see the Go icon in the chapter.

EXTENSION

CBC radioONE QUIRKS & QUARKS

Beyond the Genome

Dr. Victor Ambros (professor of genetics at Dartmouth Medical School), Dr. Katherine Wilson (associate professor of cell biology at Johns Hopkins Medical School), and Dr. Wolf Reik, (Babraham Institute in Cambridge, England) discuss their research on how our cells really work, including how genes "know" to turn on at the right times.

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Many of these questions are in the style of the Diploma Exam. You will find guidance for writing Diploma Exams in Appendix A5. Science Directing Words used in Diploma Exams are in bold type. Exam study tips and test-taking suggestions are on the Nelson Web site.

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DO NOT WRITE IN THIS TEXTBOOK.

Part 1

1. In performing experiments with fruit flies, *Drosophila melanogaster*, Thomas Morgan discovered that white eye colour is recessive to red eye colour. When females with white eyes were crossed with males with red eyes, Morgan discovered the females all had red eyes and the males all had white eyes. Select the answer that explains this outcome.

A. Male offspring inherit the white allele from the mother, which in males becomes dominant. Female offspring inherit the red allele from the father, which is dominant over the white allele they inherit from the mother.

B. Male offspring inherit the white allele from the mother and a Y chromosome from the father that does not carry a gene for eye colour. Female offspring inherit the red allele from the father, which is dominant over the white allele they inherit from the mother.

C. Male offspring inherit the red allele from the mother, which is recessive in males. Female offspring inherit the red allele from the father and no allele for eye colour from the mother.

D. Male offspring inherit the red allele from the father and a Y chromosome from the mother that carries an allele for white eye colour. Female offspring inherit the red allele from the mother, which is dominant over the white allele they inherit from the father.

Use the following information to answer questions 2 to 4.

In the pedigree chart shown in **Figure 1**, females are represented by circles and males by squares, while light shading indicates normal phenotype and dark shading indicates Duchenne muscular dystrophy.

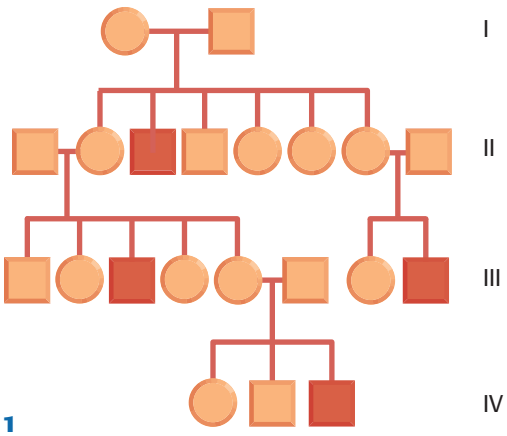

Figure 1

2. Identify the statement that correctly describes how Duchenne muscular dystrophy is inherited.

A. Duchenne muscular dystrophy is a dominant allele located on an autosome.

B. Duchenne muscular dystrophy is a recessive allele located on an autosome.

C. Duchenne muscular dystrophy is a dominant allele located on a sex chromosome.

D. Duchenne muscular dystrophy is a recessive allele located on a sex chromosome.

3. Identify the statement that is true for generation II.

A. 50 % of the males inherited the disorder from an allele carried by their mother.

B. 25 % of the males inherited the disorder from an allele carried by their mother.

C. 50 % of the females inherited the disorder from an allele carried by their father.

D. 100 % of the females inherited the allele carried by their mother but did not develop the disorder.

4. Identify the answer that is correct for generations I and III.

A. In generation I, the mother carries the recessive allele and is heterozygous. In generation III, males and females inherit the Duchenne allele from their mothers.

B. In generation I, the father carries the recessive allele and is heterozygous. In generation III, females and males inherit the Duchenne allele from their fathers.

C. In generation I, the mother carries the recessive allele and is homozygous. In generation III, only males inherit the Duchenne allele from their mothers.

D. In generation I, the father carries the recessive allele and is homozygous. In generation III, females and males inherit the Duchenne allele from their fathers.

---

5. Brown spotting on the teeth is a sex-linked trait in humans. A father with brown spotting passes the trait along to all his daughters but not to his sons. The mother does not have brown spotting on her teeth. This indicates that the brown spotting gene is

A. dominant and located on the X chromosome

B. recessive and located on the X chromosome

C. dominant and located on the Y chromosome

D. recessive and located on the Y chromosome

6. The recombination frequency among genes found on the same chromosomes depends on

A. which genes are dominant and which genes are recessive

B. the number of genes along the chromosome

C. the size of the chromosome

D. the distance between the genes

7. Ocular albinism in humans is characterized by a lack of pigment in the iris of the eyes. This X-linked trait often results in blindness for those afflicted. A woman who carries this trait marries a normal man. Identify the chance of ocular albinism in a child from this couple.
   A. 100 % chance of normal female offspring and a 100 % chance of normal male offspring
   B. 50 % chance of female offspring with ocular albinism, 50 % chance of normal female offspring, and 100 % chance of normal male offspring
   C. 100 % chance of normal female offspring, 50 % chance of male offspring with ocular albinism, and 50 % chance of normal male offspring
   D. 50 % chance of female offspring with ocular albinism, 50 % chance of normal female offspring, 50 % chance of male offspring with ocular albinism, and 50 % chance of normal male offspring

8. The allele *R* produces rose combs in chickens. Another allele *P,* located on a different chromosome, produces pea combs. The absence of the dominant rose comb and pea comb alleles (*rrpp*) produces birds with single combs. When the dominant *R* allele and the dominant *P* allele are both present, they interact to produce a walnut comb (*R_P_*). Identify the phenotypes of the parents and the expected phenotypic ratios of the F_1 generation from a cross of chickens with the genotype *RrPp* × *rrPp*.
   A. The parental phenotypes are walnut comb and pea comb. The expected F_1 phenotypic ratio from the cross is 3 walnut:3 pea:1 rose:1 single.
   B. The parental phenotypes are walnut comb and pea comb. The expected F_1 phenotypic ratio from the cross is 4 walnut:4 rose.
   C. The parental phenotypes are rose comb and pea comb. The expected F_1 phenotypic ratio from the cross is 3 walnut:2 rose:2 pea:1 single.
   D. The parental phenotypes are pea comb and single comb. The expected F_1 phenotypic ratio from the cross is 4 rose:4 pea.

Use the following information to answer questions 9 and 10.

The chromosome map in **Figure 2** shows the portion of a chromosome that carries genes for scalloped wings, bar eyes, and garnet eyes—all mutant traits in *Drosophila melanogaster*. It was drawn using data from several test crosses.

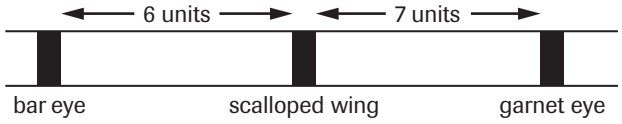

Figure 2

9. NR Determine the frequency of crossover between scalloped wings and garnet eyes, as a percent. (Record all four digits of your answer.)

10. NR Determine the frequency of crossover between bar eyes and garnet eyes, as a percent. (Record all four digits of your answer.)

Part 2

11. **Describe** Erwin Chargaff's contribution to the determination of DNA structure.

12. **Explain** how the development of the chromosome theory is linked with the development of the light microscope.

13. **Describe** the contributions made by Walter Sutton, Theodor Boveri, and Thomas Morgan in the development of the modern-day chromosome theory of genetics.

14. The gene for wild-type eye colour is dominant and sex-linked in *Drosophila melanogaster*. White eyes are recessive. The mating of a male with wild-type eye colour with a female of the same phenotype produces offspring that are $\frac{3}{4}$ wild-type eye colour and $\frac{1}{4}$ white-eyed. **Predict** the genotypes of the P_1 and F_1 generations.

15. The autosomal recessive allele *tra* transforms a female *Drosophila melanogaster* into a phenotypic male when it occurs in the homozygous condition. The transformed females are sterile. The *tra* gene has no effect on the phenotype of XY males. Using Punnett squares, **predict** the genotypes and phenotypes of individuals in the F_1 and F_2 generations from the following cross: XX, + /*tra* crossed with XY, *tra*/*tra*. (Note the + indicates the normal dominant gene.)

16. Edward Lambert, an Englishman, was born in 1717. Lambert had a skin disorder that was characterized by very thick skin, which was shed periodically. The hairs on his skin were very course and quill-like, giving him the name "porcupine man." Lambert had six sons, all of whom exhibited the same traits. The trait never appeared in his daughters. In fact, the trait has never been recorded in females. **Hypothesize** the nature of the inheritance of the "porcupine trait" that would explain these observations.

Use the following information to answer questions 17 to 20.

Figure 3 is a pedigree chart of a family in which some members have hemophilia.

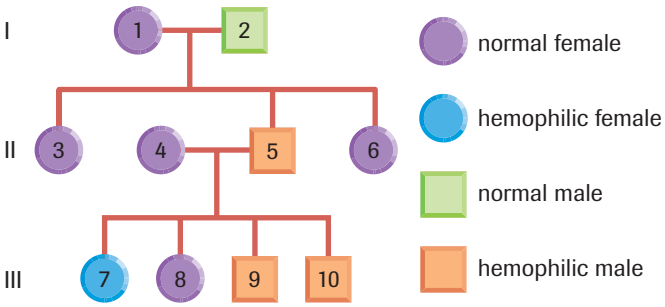

Figure 3

17. **Predict** the phenotypes of the P_1 generation.
DE

18. If parents 1 and 2 were to have a fourth child, **determine** the probability that the child would have hemophilia.
DE

19. If parents 1 and 2 were to have a second male, **determine** the probability that the boy would have hemophilia.
DE

20. **Predict** the genotypes of parents 4 and 5.
DE

21. A science student **hypothesizes** that dominant genes occur with greater frequency in human populations than recessive genes occur. Either support or refute the student's hypothesis, using the information that you have gathered in this chapter to **justify** your decision.

Use the following information to answer questions 22 to 24.

In 1911, Thomas Morgan collected the gene crossover frequencies shown in **Table 1** while studying *Drosophila melanogaster*. The loci for four different genes that code for wing shape are located on the same chromosome. Bar-shaped wings are indicated by the *B* allele, carnation wings by the *C* allele, fused veins on wings by the *FV* allele, and scalloped wings by the *S* allele.

Table 1

Gene combinations	Recombination frequency
FV/B	2.5 %
FV/C	3.0 %
B/C	5.5 %
B/S	5.5 %
FV/S	8.0 %
C/S	11.0 %

22. Use the crossover frequencies to **sketch** a gene map.
DE

23. **Identify** which genes are farthest apart. **Determine** their distance. **Illustrate** your answer by way of a diagram.
DE

24. From the data provided in **Table 1**, **conclude** in a written statement the relative position of the *FV*, *C*, and *B* alleles.
DE

chapter

20 Molecular Genetics

In this chapter

- Exploration: Similarities and Differences
- Mini Investigation: Building a DNA Model
- Web Activity: DNA Replication
- Lab Exercise 20.A: Synthesis of a Protein
- Investigation 20.1: Protein Synthesis and Inactivation of Antibiotics
- Web Activity: Electrophoresis
- Web Activity: Researchers in Human Genetic Disorders
- Mini Investigation: Examining the Human Genome
- Investigation 20.2: Restriction Enzyme Digestion of Bacteriophage DNA
- Web Activity: Transformation of Eukaryotes
- Case Study: Gene Mutations and Cancer
- Lab Exercise 20.B: Looking for SINEs of Evolution

By the mid-1950s, scientists had determined that chromosomes contained DNA and that DNA was the genetic material (**Figure 1**). Building on the work of other scientists, Watson and Crick deduced the structure of this complex molecule. This knowledge laid the basis for the field of molecular biology, which aims to understand the inheritance of traits at the level of interactions between molecules in the cell.

A primary goal of molecular genetics is to understand how DNA determines the phenotype of an organism. What happens to DNA during duplication of chromosomes in mitosis? How does the structure of DNA relate to its function? How does one molecule, identical in every somatic cell of an organism, determine the characteristics of the many different types of cells that are found in that organism?

Today, questions such as these continue to drive research in the fields of biology, biotechnology, biochemistry, and medicine. We now know the sequence of all the nucleotides that make up the genome of many organisms, including that of our own species, *Homo sapiens*. This information has given scientists new ways to study the relationships between species and the mechanisms of evolution. It also allows law enforcement agencies to identify individuals with incredible accuracy from minute quantities of DNA.

Using genetic technologies, scientists can move genes from one species to another. In fields such as agriculture, corporations have patented the genomes of these organisms in order to profit from the advantages they offer over conventional organisms. Similar manipulation of human cells may one day lead to treatments for previously untreatable debilitating diseases. The research and application of these technologies raises many social, ethical, and legal issues that society has yet to fully resolve.

STARTING Points

Answer these questions as best you can with your current knowledge. Then, using the concepts and skills you have learned, you will revise your answers at the end of the chapter.

1. Differentiate between DNA and proteins. What cellular roles do they play?
2. Describe the physical and chemical characteristics of DNA.
3. What is the significance of DNA replication in your body?
4. Write a short overview, in paragraph form, of the process of DNA replication.

Career Connections:
Biological Technician; Biotechnologist

Figure 1
DNA sequences are represented by the letters A, T, C, and G.

Exploration The Size of the Genome

All organisms, no matter how simple they may seem to us, require DNA in each cell to encode the instructions necessary to live and reproduce. The total DNA of an organism is referred to as its genome. In bacteria, the genomic DNA is circular, accounts for 2 % to 3 % of the cell's mass, and occupies about 10 % of its volume. In this activity, you will make a model of an *Escherichia coli* cell that will be 10 000 times bigger than actual size. You will also gain an appreciation for how compactly DNA is packed within a cell.

Materials: 2 cm gelatin capsule, 10 m of white thread, 10 m of coloured thread

- Try to construct the bacterium by placing the long lengths of thread inside the gelatin capsule. Good luck! It's not easy!

(a) Why does it take two lengths of thread to represent the chromosome?
(b) Is the thread that you tried to place in the capsule too thick to represent the actual thickness of the DNA? (What percentage of bacterial cell volume does your thread fill, and what is the actual volume that the DNA occupies in the bacteria?)
(c) If the human genome is 1000 times bigger than the *E. coli* genome, how many metres of thread would it take to represent the human genome?
(d) What size container would you need to hold the thread representing the human genome?

20.1 DNA Structure and Replication

According to the model proposed by Watson and Crick, DNA consists of two strands of nucleotides. Each nucleotide contains a deoxyribose sugar, a phosphate group, and a nitrogenous base, all covalently bonded to each other. Each strand of DNA has a backbone of sugar and phosphate groups (**Figure 1**). The nitrogenous bases stick out from the backbone of each DNA strand.

Watson and Crick's model also indicates that the two strands of DNA form a structure that resembles a twisted ladder. The base pairs are the rungs of the ladder and the sugar–phosphate backbones are the struts. This structure is called a double helix (see **Figure 1**). Each DNA strand in the double helix twists in a clockwise direction.

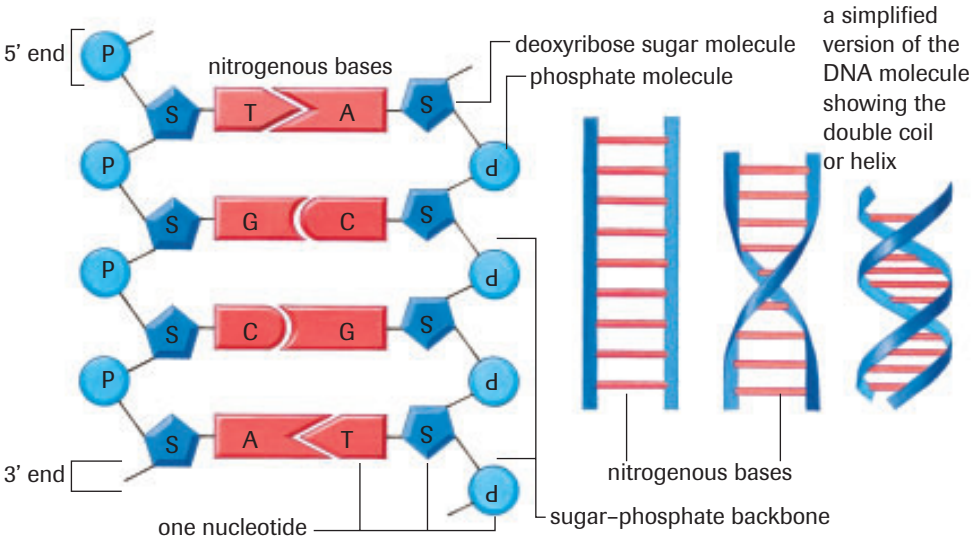

Figure 1
A simplified diagram of the structure of DNA

In the DNA molecule, the bases of one DNA strand are paired with bases in the other strand. A purine is always paired with a pyrimidine. Adenine (a purine) is always paired with thymine (a pyrimidine), and guanine (a purine) is always paired with cytosine (a pyrimidine). This type of pairing is termed **complementary base pairing**. Hydrogen bonds, between the complementary bases (A-T and G-C) on opposite strands, hold the double helix together (**Figure 2**). Although a single hydrogen bond is very weak, large numbers of hydrogen bonds are collectively strong, so the DNA molecule is very stable.

complementary base pairing
pairing of the nitrogenous base of one strand of DNA with the nitrogenous base of another strand

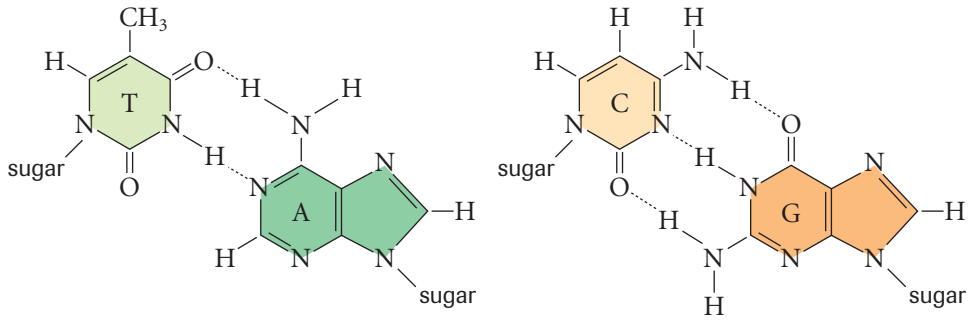

Figure 2
Adenine forms two hydrogen bonds with thymine, while guanine forms three hydrogen bonds with cytosine.

CAREER CONNECTION

Biological Technician

Biological technicians may work in the field, in the laboratory, or both. They perform routine analysis and technical duties to support the work of scientists and engineers working in fields that include molecular biology. What educational background is required to enter this field?

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The sequence of bases on any one strand of DNA can vary greatly between species, but its opposite strand will always have the complementary sequence of bases. For example, the sequences of the strands below are complementary:

5′–ATGCCGTTA–3′
3′–TACGGCAAT–5′

The two strands of nucleotides are **antiparallel**. They run parallel but in opposite directions to one another. One strand will have a 5′ carbon and phosphate group at one end and a 3′ carbon and the hydroxyl group of a deoxyribose sugar at its other end. Its antiparallel strand will have a 3′ carbon and the hydroxyl group of a deoxyribose sugar at the first end and a 5′ carbon and phosphate group at its other end (**Figure 1**, previous page).

The direction of the strand is important when enzymes interact with DNA, either to copy the DNA prior to cell division or to "read" genes in order to make proteins. Enzymes can read or copy DNA in only one direction. The sequence of only one DNA strand is given when sequences are written out since the complementary strand is easily deduced according to the rules of complementary base pairing.

antiparallel parallel but running in opposite directions; the 5′ end of one strand of DNA aligns with the 3′ end of the other strand in a double helix

Learning Tip

The rules of complementary DNA base pairing are
- A to T
- G to C

When you know the sequence on one strand, you also know the sequence on the complementary strand.

Practice

1. Define the following terms: nucleotide, complementary base pairing, and antiparallel.
2. In a DNA molecule, a purine pairs with a pyrimidine. If this is the case, then why can't A–C and G–T pairs form? (*Hint:* Look closely at the bonds between the base pairs in **Figure 2** on the previous page.)
3. The following is a segment taken from a strand of DNA: 5′–ATGCCTTA–3′. Write out the complementary strand for this segment. Be sure to show directionality.

mini Investigation — Building a DNA Model

What would a section of a DNA molecule look like if you could see one close up? You can find out by building your own model of the double helix. For this activity, you need to select materials that will allow you to model the following features:

- the sugar–phosphate backbone
- the anti-parallel strands
- the four different nitrogenous bases
- the bonds between complementary base pairs that hold the two strands together

Your model should show a minimum of 12 base pairs. It should be free-standing and approximately 15 cm tall by 6 cm wide. Include a legend with your model that clearly identifies each part of the DNA strand.

DNA Replication

In Chapter 17, you saw that mitosis involves the duplication of chromosomes. For mitosis to occur, DNA must copy itself and be equally divided between the daughter cells. To have all the correct genetic information, the DNA in each daughter cell must be an exact copy of the DNA in the parent cell. **DNA replication** is the process by which a cell makes an exact copy of its DNA. The main stages of DNA replication are the same in both prokaryotic cells (without a membrane-bound nucleus) and eukaryotic cells (with a membrane-bound nucleus).

DNA replication is semiconservative. **Semiconservative replication** involves separating the two parent strands and using them to synthesize two new strands (**Figure 3**, next page). The hydrogen bonds between complementary bases break, allowing the DNA helix to unzip. Each single DNA strand acts as a **template** to build the complementary strand. Finally, any errors are repaired, resulting in two identical DNA molecules, one for each daughter cell.

DNA replication the process whereby DNA makes exact copies of itself

semiconservative replication process of replication in which each DNA molecule is composed of one parent strand and one newly synthesized strand

template a single-stranded DNA sequence that acts as the guiding pattern for producing a complementary DNA strand

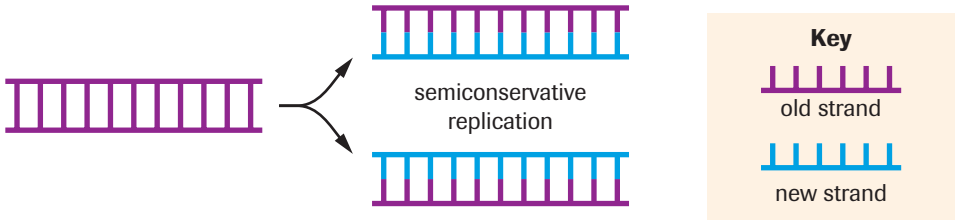

Figure 3
DNA replicates semiconservatively. Each daughter molecule receives one strand from the parent molecule plus one newly synthesized strand.

Separating the DNA Strands

DNA helicase the enzyme that unwinds double-helical DNA by disrupting hydrogen bonds

The two strands of the DNA helix cannot simply pull apart because they are tightly held together by the hydrogen bonds between bases and by the twists of the helix. The enzyme **DNA helicase** unwinds the helix by breaking the hydrogen bonds between the complementary bases. As this happens, the bonds between bases tend to reform. To prevent this, proteins bind to the separated DNA strands, helping to hold them apart. The two strands are now separated along part of the DNA molecule and are the template strands for the next step in replication. The point at which the two template strands are separating is called the replication fork. One template strand runs in the 3′ to 5′ direction in relation to the replication fork, while the other runs in the 5′ to 3′ direction (**Figure 4**).

DID YOU KNOW ?

DNA Polymerases

There are several DNA polymerases in a cell, all with their own role. Each has a unique name, created by adding a roman numeral after "DNA polymerase." The main DNA polymerase involved in DNA replication is DNA polymerase III. It adds the 5′ phosphate group of a free nucleotide to the 3′ carbon of the sugar in the last nucleotide.

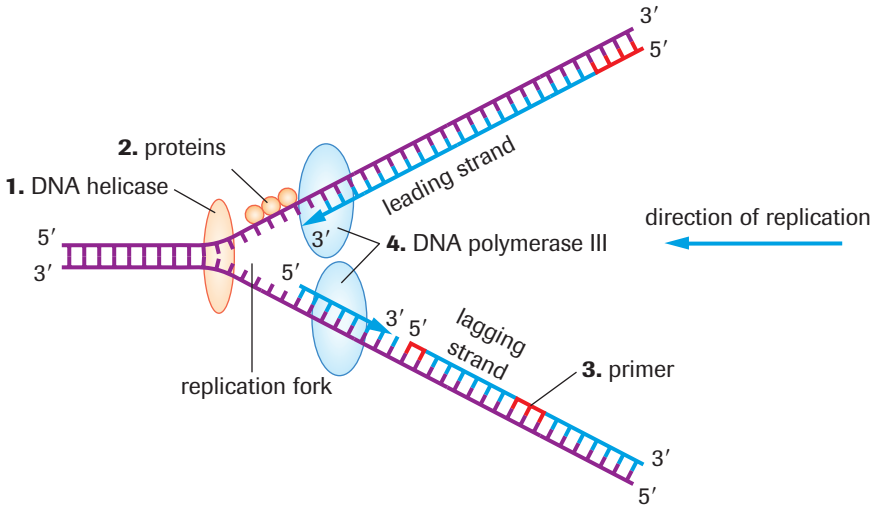

Figure 4
1. DNA helicase opens the double helix. 2. Proteins bind to the DNA to keep the two strands separate. 3. RNA primers are attached to the template strands. 4. DNA polymerase synthesizes the new DNA strands. The leading strand is synthesized continuously, and the lagging strand is synthesized in short fragments. DNA polymerase III adds complementary nucleotides in the 5′ to 3′ direction, using single-stranded primers as starting points. One nucleotide is attached to the next by bonding the phosphate on the 5′ end of the new nucleotide to the hydroxyl group on the 3′ end of the last nucleotide.

Building the Complementary Strands

DNA polymerase III the enzyme that synthesizes complementary strands of DNA during DNA replication

The next stage of DNA replication synthesizes two new DNA strands on the template strands through complementary base pairing. The new strands are synthesized by an enzyme called **DNA polymerase III**. This DNA polymerase builds a new strand by linking together free nucleotides that have bases complementary to the bases in the template. A short piece of single-stranded ribonucleic acid, called a primer, is attached to the template strand. This gives DNA polymerase III a starting point to begin synthesizing the

new DNA strand. DNA polymerase III adds nucleotides to a growing strand in *only one direction*—the 5′ to 3′ direction. The phosphate group at the 5′ end of a free nucleotide is connected to the hydroxyl group on the 3′ carbon of the sugar on the last nucleotide in the strand. As a result, one of the new strands will be synthesized continuously as DNA polymerase III moves in the 5′ to 3′ direction toward the replication fork. This strand is called the **leading strand**.

The other new strand, the **lagging strand**, is synthesized in short fragments. This allows the lagging strand to be synthesized in the 5′ to 3′ direction. RNA primers are required. To complete the replication of the DNA, the primers are cut out from the lagging strand and are replaced by DNA nucleotides by a different enzyme called **DNA polymerase I**.

Another enzyme, **DNA ligase**, links the sugar–phosphate backbone of the DNA fragments together (**Figure 5**).

leading strand the new strand of DNA that is synthesized towards the replication fork and continuously during DNA replication

lagging strand the new strand of DNA that is synthesized away from the replication fork and in short fragments, which are later joined together

DNA polymerase I an enzyme that removes RNA primers and replaces them with the appropriate nucleotides during DNA replication

DNA ligase an enzyme that joins DNA fragments together

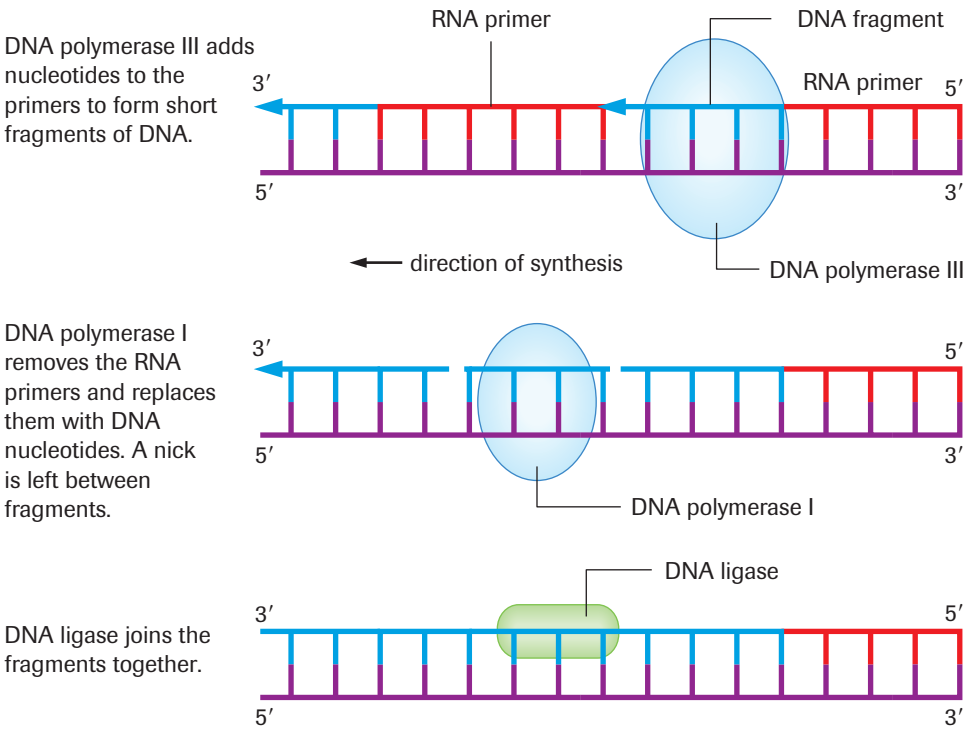

Figure 5
Building the lagging strand

DNA Repair

As complementary strands of DNA are synthesized, both DNA polymerase I and III act as quality control checkers by proofreading the newly synthesized strands. When a mistake occurs, the DNA polymerases backtrack to the incorrect nucleotide, cut it out, and then continue adding nucleotides to the complementary strand. The repair must be made immediately to avoid the mistake from being copied in later replications. Other DNA repair mechanisms can correct any errors that were missed during proofreading.

DID YOU KNOW?

Okazaki Fragments

The short fragments that are synthesized to form the lagging strand during DNA replication are called Okazaki fragments. They were named after Reiji Okazaki, who first described them in the 1960s.

WEB Activity

Simulation—DNA Replication

The *Escherichia coli* genome consists of 4.7 million nucleotide pairs. This entire genome is replicated in 40 min. Proofreading by DNA polymerase I and polymerase III maintains the error rate at roughly one error per 1000 cells duplicated! View a complete animation of DNA replication by accessing the Nelson Web site.

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SUMMARY DNA Structure and Replication

Separating the Strands

- DNA helicase unzips the double helix by breaking the hydrogen bonds between the complementary bases in the two strands of the parent DNA molecule.
- Proteins attach to the newly exposed DNA strands, preventing the hydrogen bonds from re-forming and keeping the strands apart.

Building the Complementary Strands

- DNA polymerase III adds complementary nucleotides to the growing strands, using the exposed strands of the parent DNA molecule as a template.
- The leading strand is formed continuously.
- The lagging strand is formed in short fragments, starting from an RNA primer.
- DNA polymerase I cuts out the RNA primers and replaces them with the appropriate DNA nucleotides.
- DNA ligase joins the fragments together to form a complete DNA strand.

DNA Repair

- DNA polymerase enzymes cut out incorrectly paired nucleotides and add the correct nucleotides in a process called proofreading.

Section 20.1 Questions

1. Summarize the key physical and chemical properties of DNA.
2. Differentiate between a purine and a pyrimidine.
3. Copy **Table 1** into your notebook, fill in the missing information, and supply an appropriate title.

Table 1

Enzyme	Function
DNA helicase	
DNA polymerase I	
DNA polymerase III	
DNA ligase	

4. A molecule of DNA was analyzed and found to contain 20 % thymine. Calculate the percentage of adenine, guanine, and cytosine in this molecule.
5. Define a replication fork.
6. In a double helix, there is a complete turn every 3.4 nm, or 10 nucleotides. Assume that the DNA molecule in a particular chromosome is 75 mm long. Calculate the number of nucleotide pairs in this molecule.
7. Copy **Table 2** into your notebook and complete the missing information. Explain how you determined the missing values.

Table 2

Nucleotide	Sample A	Sample B	Sample C
adenine	10 %		20 %
guanine	40 %	15 %	
thymine		35 %	20 %
cytosine			

Gene Expression 20.2

As you learned in previous chapters, specific segments of DNA on a chromosome are called genes. Genes determine the inherited characteristics, or traits, of an organism. Every somatic (body) cell in an organism contains identical copies of DNA, and each of these DNA copies is a genetic blueprint for the organism. Once scientists knew the structure of DNA and how it replicated, they used this knowledge to further investigate another question: How do the genes in DNA determine an inherited trait?

The way the information in a gene is converted into a specific characteristic or trait through the production of a polypeptide is called **gene expression**. Recall that a polypeptide is a chain of amino acids and that proteins are made up of polypeptides. Proteins form many structures in an organism, such as skin and muscle, and they also form all of the enzymes in a cell. *The products of all genes are polypeptides.*

gene expression conversion of a gene into a specific trait through the production of a particular polypeptide

A second type of nucleic acid is involved in converting the instructions in a gene into a polypeptide chain. **Ribonucleic acid (RNA)** is a polymer of nucleotides similar to DNA. There are three main structural differences between RNA and DNA. First, the sugar in RNA has an extra hydroxyl group and is called ribose rather than deoxyribose (**Figure 1**). Second, instead of the base thymine found in DNA, RNA contains the base uracil. Like thymine, uracil can form complementary base pairs with adenine (**Figure 2**). Third, RNA is single-stranded and not double-stranded like DNA. There are three types of RNA that are needed to convert genes into proteins: messenger RNA (mRNA), transfer RNA (tRNA), and ribosomal RNA (rRNA).

ribonucleic acid (RNA) a nucleic acid consisting of nucleotides comprised of the sugar ribose and nitrogenous bases

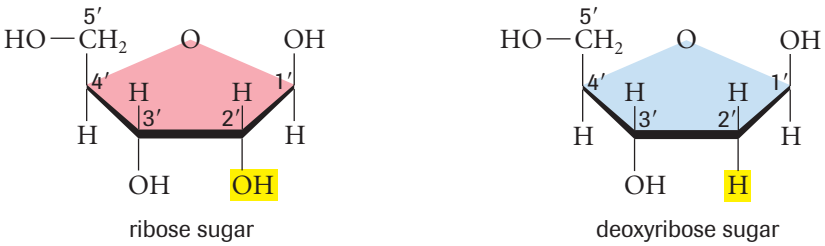

Figure 1
A ribose sugar possesses an —OH group (hydroxyl) on the 2′ carbon. The deoxyribose sugar is missing the —OH group on the 2′ carbon. The *deoxy* part of the name deoxyribose indicates a "loss of oxygen" at position 2.

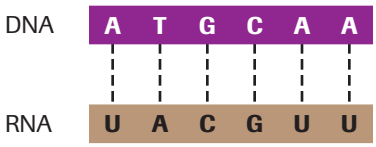

Figure 2
Base pairing of RNA with DNA during transcription. Notice that thymine does not exist in RNA but is substituted with uracil.

The Central Dogma

There are two main stages of gene expression, transcription and translation. In **transcription**, the genetic information is converted from a DNA sequence into **messenger RNA (mRNA)**. In all cells, the mRNA carries the genetic information from the chromosome to the site of protein synthesis. In eukaryotic cells, which contain a nucleus, the mRNA carries the genetic information from the nucleus to the cytoplasm as it passes through the pores in the nuclear envelope.

transcription the process of converting DNA into messenger RNA

messenger RNA (mRNA) the product of transcription of a gene; mRNA is translated by ribosomes into protein

The second stage of gene expression is **translation**. During translation, the genetic information carried by the mRNA is used to synthesize a polypeptide chain.

translation the process of synthesizing a specific polypeptide as coded for by messenger RNA

The two-step process of transferring genetic information from DNA to RNA and then from RNA to protein is known as the central dogma of molecular genetics (**Figure 3**, next page). We will explore transcription and translation in more detail in this section. You

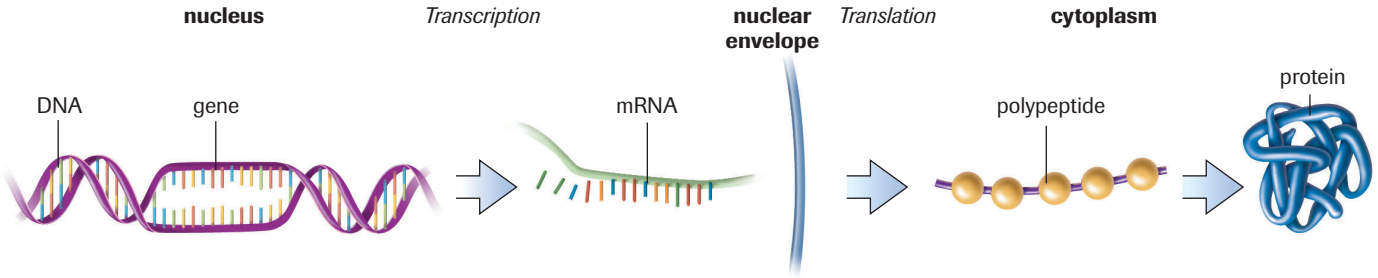

Figure 3
The central dogma of molecular genetics

will see that the sequence of nucleotides in a gene determines the sequence of amino acids in a polypeptide.

Transcription

During transcription, the DNA sequence of a gene is copied (transcribed) into the sequence of a single-stranded mRNA molecule.

Transcription is divided into three processes: initiation, elongation, and termination. During initiation, an enzyme called **RNA polymerase** binds to the DNA at a specific site near the beginning of the gene. During elongation, RNA polymerase uses the DNA as a template to build the mRNA molecule. During termination, the RNA polymerase passes the end of the gene and comes to a stop. The mRNA is then released from the template strand of DNA.

RNA polymerase enzyme that transcribes DNA

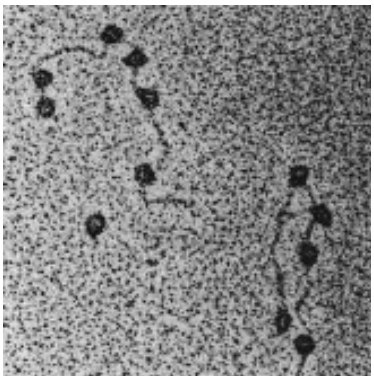

Figure 4
The RNA polymerase (dark circles) binds to the DNA strand and initiates transcription. Transcription occurs simultaneously at numerous locations along the DNA.

Initiation

Transcription starts when the RNA polymerase enzyme binds to the segment of DNA to be transcribed and opens the double helix. **Figure 4** shows an electron micrograph of this process. The RNA polymerase binds to the DNA molecule in front of the gene to be transcribed in a region called the **promoter**. In most genes, the promoter sequence contains a string of adenine and thymine bases that serves as the recognition site for RNA polymerase. The promoter indicates which DNA strand should be transcribed and where the RNA polymerase should start transcribing the DNA. Since the binding site of RNA polymerase only recognizes the promoter region, it can only bind in front of a gene.

promoter sequence of DNA that binds RNA polymerase in front of a gene

Elongation

Once the RNA polymerase binds to the promoter and opens the double helix, it starts building the single-stranded mRNA in the 5′ to 3′ direction. The promoter is not transcribed. The process of elongation of the mRNA molecule is similar to DNA replication. However, RNA polymerase does not require a primer and it copies only one of the DNA strands. The transcribed DNA strand is called the **template strand**. The mRNA sequence is complementary to the DNA template strand except that it contains the base uracil in place of thymine.

template strand the strand of DNA that the RNA polymerase uses as a guide to build complementary mRNA

Termination

Synthesis of the mRNA continues until RNA polymerase reaches the end of the gene. RNA polymerase recognizes the end of the gene when it comes to a stop signal called a **termination sequence**. Transcription stops and the newly synthesized mRNA disconnects from the DNA template strand. RNA polymerase is then free to bind to another promoter region and transcribe another gene. **Figure 5**, on the next page, summarizes the steps in transcription.

termination sequence sequence of bases at the end of a gene that signals the RNA polymerase to stop transcribing

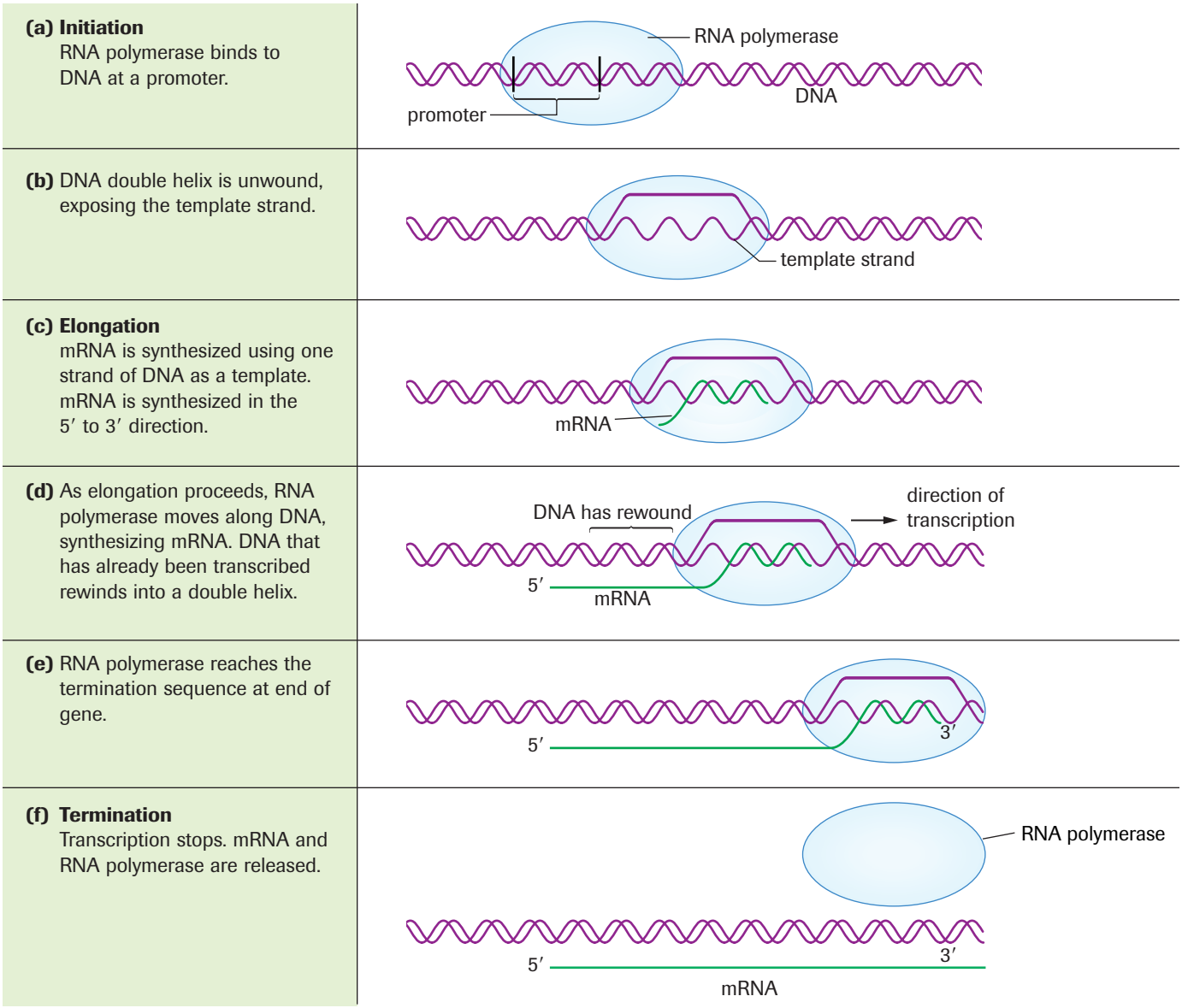

Figure 5
A summary of the process of transcription

Practice

1. A short fragment of a particular gene includes the following sequence of nucleotides:

   TACTACGGT

   Write out the corresponding mRNA transcript.

2. A short fragment of another gene includes the following sequence of nucleotides:

   ACCATAATATTACCGACCT TCG

   (a) Explain the purpose of the promoter region in transcription.
   (b) Copy the sequence into your notebook and circle the promoter region. Explain the rationale for your selection.

EXTENSION

Regulation of Transcription
This Audio Clip discusses the regulatory factors that control when and how much mRNA is transcribed from a given gene.

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Translation

The second part of the central dogma of molecular biology (**Figure 3**, page 668) is the translation of the genetic information carried by mRNA into a chain of amino acids to form a polypeptide. Therefore, the process of translation involves protein synthesis, and it depends on the remarkable nature of the genetic code.

Only 20 amino acids are found in proteins. The DNA in a gene codes for these 20 amino acids by combinations of the four nitrogenous bases. During translation, the DNA code is read in groups of three nucleotides, called a **codon**. Each codon calls for a specific amino acid to be placed in the growing polypeptide chain. Codons can consist of any combination of the four nitrogenous bases, so there are 64 ($4^3 = 64$) possible different codons for the 20 different amino acids. The groups of three bases in both DNA and mRNA are both called codons, so it is important to clarify which code is being presented when writing out a genetic sequence. The remainder of this description will use mRNA codons. **Table 1** shows the mRNA codons. One of these codons (AUG) is the **start codon**, where translation begins. It also codes for the insertion of the amino acid methionine, so all polypeptide chains initially start with the methionine, but it may later be edited out. Three other codons (UAA, UAG, and UGA) do not code for amino acids and are called the **stop codons** because they cause protein synthesis to stop. The other 60 codons code for one of the 20 amino acids. Some amino acids have more than one codon; for example, both serine and leucine each have 6 different codons. **Table 2**, on the next page, lists the abbreviations for the amino acids to help you look them up in **Table 1**.

codon sequence of three bases in DNA or complementary mRNA that serves as a code for a particular amino acid

start codon specific codon (AUG) that signals the start of translation

stop codon specific codon that signals the end of translation

Like transcription, translation can be divided into the same three stages: initiation, elongation, and termination.

Table 1 Codons and Their Amino Acids

1st Base	2nd (middle) Base of a Codon				3rd Base
	U	C	A	G	
U	UUU Phe	UCU Ser	UAU Tyr	UGU Cys	U
	UUC Phe	UCC Ser	UAC Tyr	UGC Cys	C
	UUA Leu	UCA Ser	UAA STOP	UGA STOP	A
	UUG Leu	UCG Ser	UAG STOP	UGG Trp	S
C	CUU Leu	CCU Pro	CAU His	CGU Arg	U
	CUC Leu	CCC Pro	CAC His	CGC Arg	C
	CUA Leu	CCA Pro	CAA Gln	CGA Arg	A
	CUG Leu	CCG Pro	CAG Gln	CGG Arg	S
A	AUU Ile	ACU Thr	AAU Asn	AGU Ser	U
	AUC Ile	ACC Thr	AAC Asn	AGC Ser	C
	AUA Ile	ACA Thr	AAA Lys	AGA Arg	A
	AUG Met	ACG Thr	AAG Lys	AGG Arg	S
G	GUU Val	GCU Ala	GAU Asp	GGU Gly	U
	GUC Val	GCC Ala	GAC Asp	GGC Gly	C
	GUA Val	GCA Ala	GAA Glu	GGA Gly	A
	GUG Val	GCG Ala	GAG Glu	GGG Gly	S

EXTENSION

Why Three Nucleotides per Codon?

Why are there always three nucleotides in a codon? Why not two or four? Listen to this Audio Clip to find out the reason behind the triplet code found in DNA and mRNA sequences.

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Initiation

Initiation of translation occurs when a **ribosome** recognizes a specific sequence on the mRNA and binds to that site. In eukaryotes, the ribosome consists of two subunits, a large subunit and a small subunit (**Figure 6**, next page). The two subunits bind to the mRNA, clamping it between them. The ribosome then moves along the mRNA in the 5′ to 3′ direction, adding a new amino acid to the growing polypeptide chain each time it

ribosome an organelle composed of RNA and protein and located in the cytoplasm that carries out protein synthesis

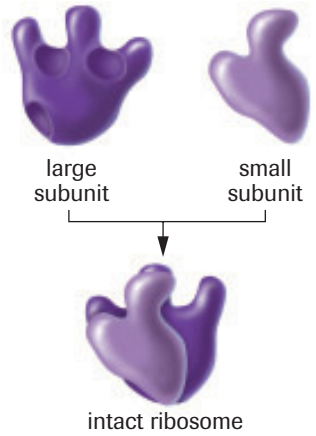

Figure 6
Ribosomes consist of a large subunit and a small subunit.

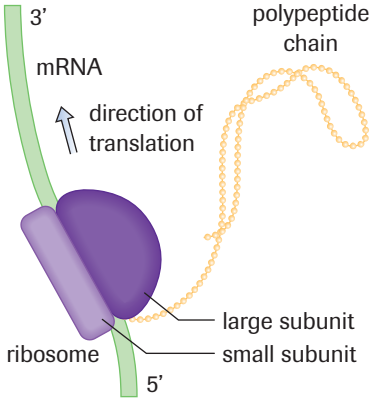

Figure 7
The large and small subunit of a ribosome work together to translate a strand of mRNA into a polypeptide. The polypeptide grows as the ribosome moves farther along the mRNA strand.

reads a codon (**Figure 7**). Ribosomes synthesize different proteins by associating with different mRNAs and reading their coding sequences.

A ribosome must begin reading the coding sequence at the correct place in the mRNA, or it will misread all the codons. The first codon that it recognizes is the start codon AUG. Binding to the start codon ensures that the ribosome translates the genetic code using the reading frame of the mRNA molecule. It is critical that the mRNA be positioned in the ribosome in its reading frame so that the genetic code is translated into the correct sequence of amino acids.

Once the ribosome has bound the mRNA, how does it get the amino acids that correspond to the codon? This job falls to a second type of RNA molecule known as **transfer RNA (tRNA)**. At one end of the tRNA there is a sequence of three bases, the **anticodon**, that is complementary to the codon of the mRNA. The opposite end carries the corresponding amino acid (**Figure 8**, next page). For example, if the mRNA has the codon UAU, the complementary base sequence of the anticodon is AUA, and the tRNA would carry the amino acid tyrosine. Check **Table 1** to find the mRNA codon and prove to yourself that it calls for tyrosine. Every tRNA carries only one specific amino acid, which means that at least 20 different tRNAs are required. Recall that there are 64 possible codons. In reality, anywhere from 20 to 64 types of tRNA molecules are available, depending on the organism.

Table 2 Amino Acids and Their Abbreviations

Amino acid	Three-letter abbreviation
alanine	Ala
arginine	Arg
asparagine	Asn
aspartic acid	Asp
cysteine	Cys
glutamic acid	Glu
glutamine	Gln
glycine	Gly
histidine	His
isoleucine	Ile
leucine	Leu
lysine	Lys
methionine	Met
phenylalanine	Phe
proline	Pro
serine	Ser
threonine	Thr
tryptophan	Trp
tyrosine	Tyr
valine	Val

transfer RNA (tRNA) the form of RNA that delivers amino acids to a ribosome during translation

anticodon group of three complementary bases on tRNA that recognizes and pairs with a codon on the mRNA

Practice

3. Transcribe the following sequence of DNA into mRNA.
   TACGGATTTCTCCGCAAATTAGGG
4. Translate the following mRNA sequence into an amino acid sequence.
   5′-AUGCCCUCUAUUCCGGGAAGAUAG-3′
5. How many nucleotides are necessary in the DNA to code for the following sequence of amino acids?
   Leu-Tyr-Arg-Trp-Ser

DID YOU KNOW

RNA Polymerase I, II, III

There are three forms of the RNA polymerase in eukaryotes: RNA polymerase I transcribes ribosomal RNA; RNA polymerase II transcribes mRNA; and RNA polymerase III transcribes tRNA and other short genes that are about 100 base pairs in length.

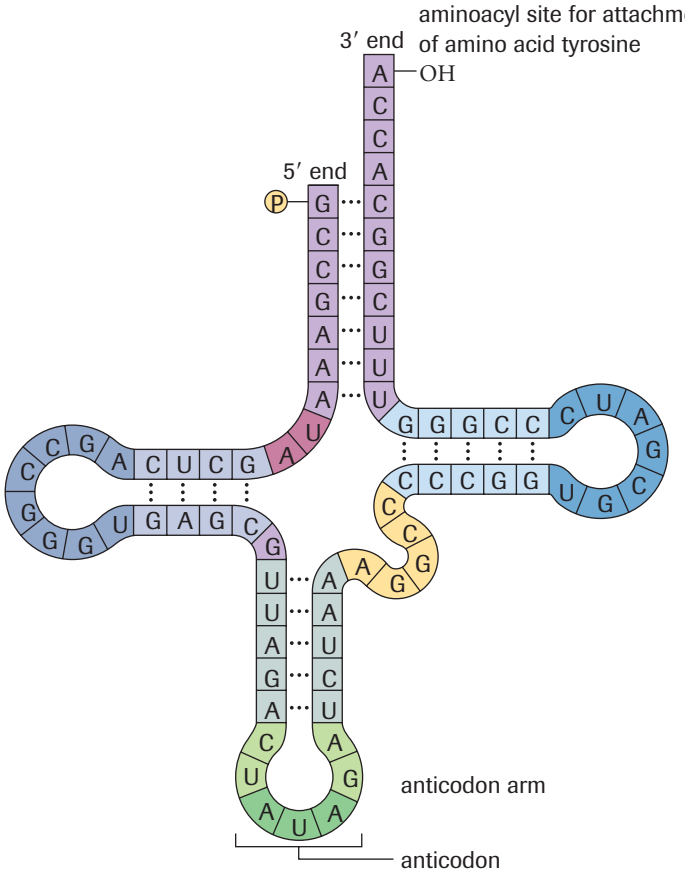

Figure 8

The tRNA molecule has a cloverleaf structure. The molecule folds to form this structure because of hydrogen bonding. The anticodon is located on the anticodon arm and the amino acid is covalently bound to the adenine nucleotide at the 3′ end (aminoacyl). In this case, the amino acid that would be added is tyrosine because the anticodon is AUA.

Elongation

The first codon that is recognized by the ribosome is the start codon AUG. The AUG codon also codes for methionine, so every protein initially starts with the amino acid methionine. The ribosome has two sites for tRNA to attach: the A (aminoacyl) site and the P (peptidyl) site. The tRNA with the anticodon complementary to the start codon enters the P site, as shown in **Figure 9** (**a**). The next tRNA carrying the required amino acid enters the A site, as shown in **Figure 9** (**b**). In **Figure 9** (**c**), a peptide bond has formed between the methionine and the second amino acid, alanine. The ribosome has shifted over one codon so that the second tRNA is now in the P site. This action has released the methionine-carrying tRNA from the ribosome and allowed a third tRNA to enter the empty A site. The process is similar to a tickertape running through a tickertape machine, except that the ribosome "machine" moves along the mRNA "tickertape." The tRNAs that have been released are recycled in the cell cytoplasm by adding new amino acids to them. The process continues until the entire code of the mRNA has been translated and the ribosome reaches a stop codon, as shown in **Figures 9** (**d**) and (**e**).

DID YOU KNOW

From DNA to Protein

The discovery of the relationship between DNA, mRNA, ribosomes, tRNA, and protein was the result of numerous scientists working on separate pieces of the puzzle. Watch an online animation of their studies.

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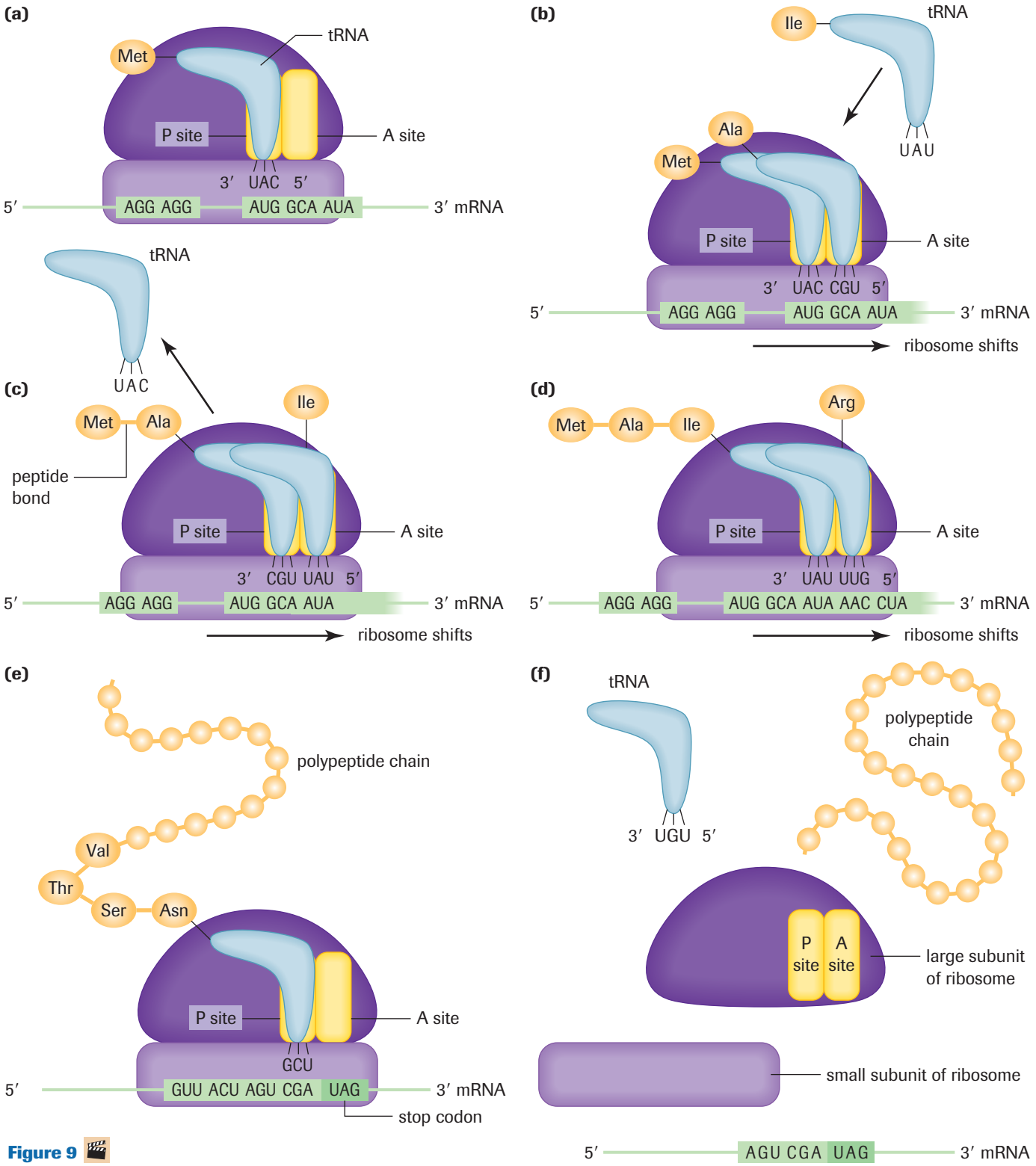

Figure 9
Protein synthesis

(a) The first tRNA that is brought into the P site carries methionine because the start codon is AUG.
(b) The second tRNA enters the A site.
(c) A peptide bond forms between methionine and alanine. The ribosome shifts one codon over and the next tRNA brings in the appropriate amino acid into the A site.
(d) The ribosome moves the mRNA and another amino acid is added to the chain.
(e) The process is repeated until the ribosome reaches a stop codon for which no tRNA exists.
(f) A release-factor protein helps break apart the ribosome–mRNA complex, releasing the polypeptide chain.

Termination

Eventually, the ribosome reaches one of the three stop codons: UGA, UAG, or UAA. Since these three codons do not code for an amino acid, there are no corresponding tRNAs. A protein known as a release factor recognizes that the ribosome has stalled and helps release the polypeptide chain from the ribosome. As shown in **Figure 9 (f)**, on the previous page, the two subunits of the ribosome now fall off the mRNA and translation stops.

LAB EXERCISE 20.A

Report Checklist

○ Purpose	○ Design	● Analysis
○ Problem	○ Materials	○ Evaluation
○ Hypothesis	○ Procedure	○ Synthesis
○ Prediction	○ Evidence	

Synthesis of a Protein

In this activity, you are provided with a DNA nucleotide sequence that codes for a hypothetical protein. The code is given in three fragments. This DNA code is from a eukaryotic cell so in the mRNA transcript there are extra codons called introns. Eukaryotic cells cut these sequences out of the mRNA before it leaves the nucleus, so the codons are transcribed but are not translated.

In this exercise, you will transcribe the three pieces of DNA code into mRNA and identify the beginning fragment, the middle fragment, and the end fragment. In addition, you will remove the intron segment and translate the mRNA into the protein.

Procedure

1. Copy each of the following sequences onto a separate piece of paper. (*Hint:* Turn your paper so you can write the sequence out along the horizontal length of the paper. Leave room below each sequence to write your mRNA sequence directly below.)

   Sequence A
   CTCGCGCCGAAACTTCCCTCCTAAACGTTCAAC
   CGGTTCTTAATCCGCCGCCAGGGCCCC

   Sequence B
   CGTAACAACTTGTTACAACATGGTCATAAACGTCA
   GATGGTCAATCTCTTAATGACT

   Sequence C
   TACAAACATGTAAACACACCCTCAGTGGACCAA
   CTCCGCAACATAAACCAAACACCG

2. Divide the sequences into triplets (codons) by putting a slash between each group of three bases.
3. Transcribe the DNA into mRNA.
4. Identify the middle, end, and beginning sequence. Use your knowledge of start and stop codons to help you figure it out. (*Hint:* You will need to examine the codons that start and end a fragment.)
5. Remove codons 24 to 51, including codon 51. These codons are the intron, or extra codons, found in this DNA segment.
6. Translate the mRNA into protein using the genetic code.

Analysis

(a) Which fragment was the beginning fragment? How do you know?

(b) Which fragment was the end fragment? How do you know?

(c) Codons 24 to 51 represent an intron. If the introns were not cut out of the mRNA before it leaves the nucleus and attaches to a ribosome, what would happen to the protein structure? Is it likely that this protein would still perform the same function? Explain your answer.

(d) How many amino acids does this protein contain?

(e) Is this genetic sequence eukaryotic or prokaryotic? How do you know?

(f) If you worked backward, starting with the amino acid sequence of the protein, would you obtain the same DNA nucleotide sequence? Why or why not?

(g) Provide the anticodon sequence that would build this protein.

INVESTIGATION 20.1 *Introduction*

Protein Synthesis and Inactivation of Antibiotics

Each protein has a specific function. Its presence or absence in a cell may make the difference between life and death. Bacteria that carry an ampicillin-resistance gene produce a protein that inactivates the antibiotic ampicillin. What happens when they are grown on ampicillin-rich media? This investigation allows you to observe the effects of the presence and function of a specific gene.

Report Checklist

● Purpose	● Design	● Analysis
○ Problem	○ Materials	● Evaluation
● Hypothesis	○ Procedure	● Synthesis
● Prediction	● Evidence	

To perform this investigation, turn to page 695.

SUMMARY *Gene Expression*

Table 3 Summary of Transcription

Initiation
• Initiation of transcription starts when the RNA polymerase binds to the promoter region of the gene to be transcribed.
• The DNA is unwound and the double helix is disrupted.
Elongation
• A complementary messenger RNA (mRNA) molecule is synthesized in the 5′ to 3′ direction, using one strand of DNA as a template.
• Adenine (A) bases in the DNA are paired with uracil (U) in the mRNA.
• Transcription continues until the RNA polymerase reaches a termination sequence.
Termination
• When the RNA polymerase comes to a termination sequence, it falls off the DNA molecule.
• The mRNA separates from the DNA.

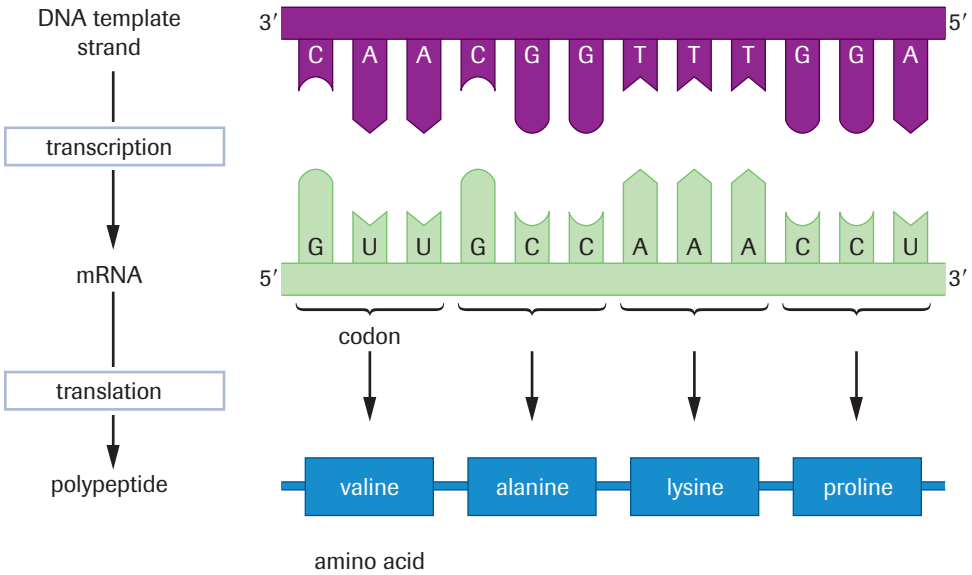

Figure 10
An overview of gene expression

Table 4 Summary of Translation

Initiation
• Ribosome subunits (large and small) bind to the mRNA transcript, sandwiching the mRNA between them. • The ribosome moves along the mRNA, reading the codons. • Translation begins when the ribosome reaches the start codon, AUG.
Elongation
• Through the genetic code, each codon specifies a particular one of the 20 amino acids that make up polypeptides. • Transfer RNA (tRNA) molecules have an anticodon that is complementary to the codon in the mRNA. The tRNA carries the amino acid specified by the codon. • The ribosome contains two sites, the A (aminoacyl) site and the P (peptidyl) site. • When the start codon is in the P site, the first tRNA delivers methionine. Since the start codon codes for methionine, all polypeptides initially start with this amino acid. • The second codon of the mRNA is exposed at the same time in the A site. When the tRNA delivers the second amino acid, a peptide bond is formed between the two amino acids. • The ribosome shifts over one codon. The tRNA that delivered the methionine is released from the P site. • When the ribosome shifts, the tRNA containing the growing polypeptide moves to the P site. A third amino acid, specified by the third codon, is brought in to the A site by the next tRNA. A peptide bond is formed between the second and third amino acid. • Amino acids continue to be added to the polypeptide until a stop codon is read in the A site.
Termination
• The stop codons are UAG, UGA, and UAA. At this point the ribosome stalls. • A protein known as the release factor recognizes that the ribosome has stalled and causes the ribosome subunits to disassemble, releasing the mRNA and newly formed polypeptide.

Section 20.2 Questions

1. State the central dogma of molecular genetics.
2. Describe the role of the following molecules in gene expression: ribosomes, mRNA, tRNA.
3. The genetic code is read in groups of three nucleotides called codons. Explain why reading the code in pairs of nucleotides is not sufficient.
4. The following is the sequence of a fragment of DNA:

   GGATCAGGTCCATAC

   Transcribe this sequence into mRNA.
5. Using the genetic code, decipher the following mRNA sequence:

   5′ - AUGGGACAUUAUUUUGCCCGUUGUGGU - 3′
6. The amino acid sequence for a certain peptide is Leu–Tyr–Arg–Trp–Ser. How many nucleotides are necessary in the DNA to code for this peptide?
7. Identify which step in transcription would be affected and predict what would happen in each situation:
   (a) The termination sequence of a gene is removed.
   (b) RNA polymerase fails to recognize the promoter.
8. Construct a table to compare the processes of replication and transcription. Remember to consider both similarities and differences.
9. Distinguish between the following terms:
   (a) P site and A site
   (b) codon and anticodon
   (c) start and stop codon
   (d) DNA and RNA
10. Identify which of the following selections correctly lists the anticodons for the amino acids threonine, alanine, and proline:

A.	ACU	GCU	CCA
B.	ACT	GCT	CCA
C.	TGA	CGA	GGT
D.	UGA	CGA	GGU

11. Errors are occasionally made during the process of transcription. Explain why these errors do not always result in an incorrect sequence of amino acids. Describe at least two examples to illustrate your answer.

DNA and Biotechnology 20.3

Carpenters require tools such as hammers, screwdrivers, and saws, and surgeons require scalpels, forceps, and stitching needles. The tools of the molecular biologist are living biological organisms or biological molecules. Using these tools, scientists can treat specific DNA sequences as modules and move them from one DNA molecule to another, forming **recombinant DNA**. Research in exploring and using this type of biotechnology has led to exciting new advances in biological, agricultural, and medical technology. Biotechnology research has also found ways to introduce specific DNA sequences into a living cell. For example, the gene that encodes insulin has been introduced into bacterial cells so that they become living factories producing this vital hormone. The introduction and expression of foreign DNA in an organism is called **genetic transformation**. In this section, you will explore some of the key tools used by molecular geneticists in producing recombinant DNA and genetically transformed organisms.

recombinant DNA fragment of DNA composed of sequences originating from at least two different sources

genetic transformation introduction and expression of foreign DNA in a living organism

DNA Sequencing

Before a DNA sequence can be used to make recombinant DNA or to transform an organism, the scientist or technician must first identify and isolate a piece of DNA containing that sequence. One of the tools used to do this is DNA sequencing. DNA sequencing determines the exact sequence of base pairs for a particular DNA fragment or molecule. In 1975, the first DNA sequencing techniques were simultaneously developed by Frederick Sanger and his colleagues and by Alan Maxim and Walter Gilbert. Sanger's technique relied on first replicating short segments of DNA that terminate due to a chain-terminating nucleotide. Four separate reaction tubes are run, each with a chain-terminating nucleotide incorporating a different base (i.e., A, T, G, and C). The various lengths of DNA segments are then separated by loading and running the contents of the tubes on a sequencing gel (**Figure 1**). Because the end nucleotide of each segment is chain-terminating, its base is already known. Consequently, the sequence can be read directly from the gel in ascending order (shortest to longest segments). The sequence of the strand is written along the edge of the gel diagram, starting from the bottom where the shortest strands have travelled. This method is comparatively slow and can only sequence short fragments of DNA.

DNA can also be sequenced in a test tube using isolated segments of DNA. This technique depends on a primer, DNA polymerase, and the four DNA nucleotides, each of which is labelled with a specific dye. The complementary strand is built from these dye-labelled nucleotides. The nucleotides in the synthesized strand can then be identified by their colours, allowing the original strand sequence to be deduced according to the rules of complementary base pairing.

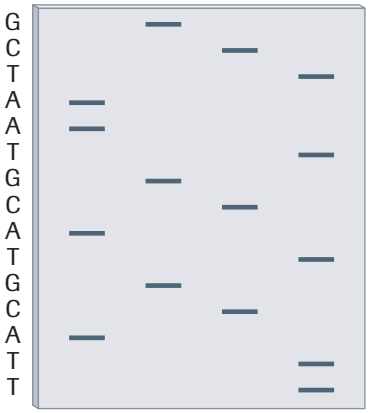

Figure 1
A sequencing gel is a matrix containing many small spaces. The DNA fragments are charged and will move towards one pole of an electric field. Smaller DNA fragments move through the spaces more quickly than larger fragments and are found at the bottom of the gel. The larger fragments will remain towards the top of the gel. The resulting ladder of fragments can be read, giving the sequence of the initial DNA fragment.

WWW WEB Activity

Simulation—Electrophoresis

Electrophoresis is an important tool in molecular biology. In addition to nucleic acids, it is also used to separate proteins from a mixture. Electrophoresis of nucleic acids and proteins depend on the similar factors. In this Virtual Biology Lab, you will perform polyacrylamide gel electrophoresis (PAGE) to identify proteins involved in the biochemistry of shell colour in an extinct organism.

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Figure 2
Dr. Judith Hall

WEB Activity

Canadian Achievers—Researchers in Human Genetic Disorders

Advances in biotechnology have led a greater understanding of many human genetic disorders. These advances have involved many research teams working together, either directly or by publishing their work in peer-reviewed articles. The following list shows some Canadians who are among the researchers making important contributions:

- Dr. Michael Hayden, University of British Columbia: Huntington disease
- Dr. Lap-Chee Tsui, Hospital for Sick Children, Toronto: cystic fibrosis
- Dr. Judith Hall, University of British Columbia: cystic fibrosis
- Dr. Christine Bear, University of Toronto: cystic fibrosis
- Dr. Ron Warton, Hospital for Sick Children, Toronto: Duchenne muscular dystrophy

Go to the Nelson Web site to find more information on the work of these people. After you have completed reading this material, write a short paragraph that describes your view on the importance of genetic research. Defend your position.

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DID YOU KNOW

Genome Facts

On February 15, 2001, scientists from the Human Genome Project and Celera Genomics confirmed that there were approximately 30 000 genes in the human genome—a number far less than the original estimate of 120 000. This was determined using two different DNA sequencing techniques.

Other Facts

- 99.9 % of the nitrogenous base sequences is the same in all humans.
- Only 5 % of the genes contains the instructions for producing functional proteins; the remaining 95 % does not have any known function.
- A worm has approximately 18 000 genes; a yeast cell has about 6000.

The Human Genome Project

In a series of meetings held in the mid-1980s, plans were developed to begin the process of producing maps of the entire genetic makeup of a human being. The international project began in the United States in October 1990 with James Watson, of Nobel Prize fame, as one of the first directors. The human genome consists of approximately 30 000 genes, with the 23 pairs of chromosomes containing an estimated 3 billion pairs of nucleotides. Constructing the genome map involved using mapping techniques (similar to those you read about in Chapter 19) and DNA sequencing technology. When the project began, only about 4500 genes had been identified and sequenced. The collaborative efforts of many scientists from numerous countries and rapid improvements in sequencing techniques helped complete the gene map by June 2000 (**Table 1**).

Table 1 Milestones in Genome Mapping

Milestone	Date
human chromosome 22 completed (the first chromosome to be mapped)	December 1999
Drosophila genome completed	March 2000
human chromosomes 5, 16, 19 mapped	April 2000
human chromosome 21 completely mapped	May 2000
human genome completely mapped	June 2000

A DNA sequencing technique based on the one developed by Sanger was the most common method used in the project. In this technique, pieces of DNA are replicated and changed so that the fragments, each ending with one of the four nucleotides, can be detected by a laser. Automated equipment can then determine the exact number of nucleotides in the chain. A computer is used to combine the huge amount of data and reconstruct the original DNA sequence.

Prior to the Human Genome Project, the genes for hereditary disorders such as cystic fibrosis, muscular dystrophy, and Huntington disease had been identified. The aim of the project is to add to this list so that new drugs and genetic therapies can be developed to

mini Investigation — Examining the Human Genome

In this activity, you will go to an online map of the human genome. On the map, you will find diagrams containing information about every chromosome in the genome. The magenta and green regions on the diagrams reflect the unique patterns of light and dark bands seen on human chromosomes that have been stained for viewing through a light microscope. The red region represents the centromere or constricted portion of the chromosome. On other chromosome diagrams, you will see yellow regions that mark chromosomal areas that vary in staining intensity. The chromatin in these areas is condensed and sometimes known as heterochromatin, meaning "different colour." Some diagrams have yellow regions overlaid by thin horizontal magenta lines. This colour pattern indicates variable regions called stalks that connect very small chromosome arms (satellites) to the chromosome.

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Go to the Nelson Web site, and follow the link for Mini Investigation: Examining the Human Genome. On the genome map, click on each chromosome diagram to discover the traits and disorders located on that chromosome. For example, **Figure 3** shows traits and disorders that are found on chromosome 20.

Touch each chromosome pair to find the number of genes mapped on that chromosome.

Use the information you find to answer the questions below.

(a) Which chromosome pair contains the greatest number of genes?

(b) Which chromosome contains the fewest genes?

(c) Estimate the size of the human genome. Show how you calculated your estimate.

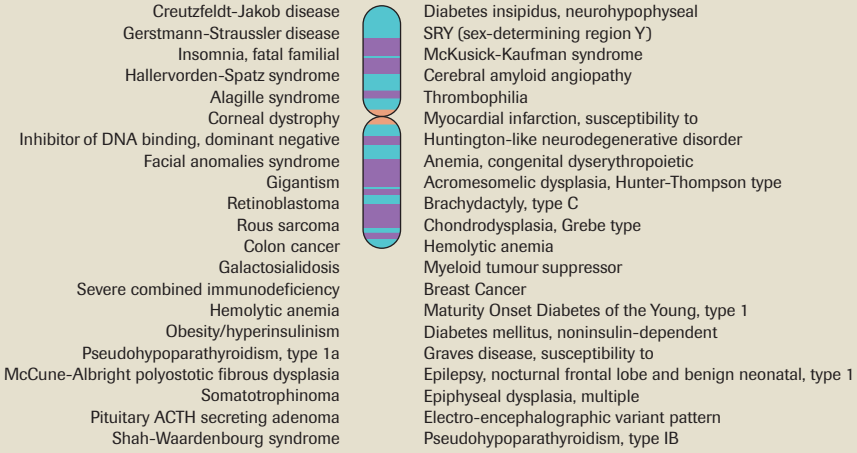

Figure 3
Although chromosome 20 is one of the smallest chromosomes, it has a great number of genes.

combat genetic disorders. The project also may open a Pandora's box of ethical questions, legal dilemmas, and societal problems. Who will own or control the information obtained and how will we prevent potential misuse of the data?

Enzymes and Recombinant DNA

As you have seen in this section, DNA sequencing is one way to identify specific segments of DNA. Another way is by creating genetic linkage maps, as you saw in Chapter 18. Once a particular segment of DNA has been identified, molecular biologists may use enzymes to isolate that segment or modify it. The DNA fragment may then be used to create recombinant DNA or be transferred to another organism. We will review some of the most commonly used enzymes.

CAREER CONNECTION

Biotechnologist

Biotechnologists are involved in improving and developing processes and products used in agriculture, health care, and the chemical industry. A biotechnologist needs specialized knowledge of biochemistry, microbiology, and molecular genetics. Find out more about opportunities in this field.

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Restriction Endonucleases

restriction endonuclease an enzyme that cuts double-stranded DNA into fragments at a specific sequence; also known as a restriction enzyme

recognition site a specific sequence within double-stranded DNA that a restriction endonuclease recognizes and cuts

palindromic reading the same backwards and forwards

Restriction endonucleases, otherwise known as restriction enzymes, are like molecular scissors that can cut double-stranded DNA at a specific base-pair sequence. Each type of restriction enzyme recognizes a particular sequence of nucleotides that is known as its **recognition site**. Molecular biologists use these enzymes to cut DNA in a predictable and precise way. Most recognition sites are four to eight base pairs long and are usually characterized by a complementary **palindromic** sequence (**Table 2**). For example, look at the restriction enzyme *Eco*RI. This sequence is palindromic because both strands have the same base sequence when read in the 5′ to 3′ direction.

Table 2 Restriction Enzymes and Their Recognition Sites

Microorganism of origin	Enzyme	Recognition site	After restriction enzyme digestion
Escherichia coli	*Eco*RI	5′-G↓AATTC-3′ 3′-CTTAA↑G-5′	5′-G 3′-CTTAA AATTC-3′ G-5′
Serratia marcescens	*Sma*I	5′-CCC↓GGG-3′ 3′-GGG↑CCC-5′	5′-GGG 3′-CCC CCC-3′ GGG-5′
Arthrobacter luteus	*Alu*I	5′-AG↓CT-3′ 3′-TC↑GA-5′	5′-AG 3′-TC CT-3′ GA-5′
Streptomyces albus	*Sal*I	5′-G↓TCGAC-3′ 3′-CAGCT↑G-5′	5′-G 3′-CAGCT TCGAC-3′ G-5′
Haemophilus parainfluenzae	*Hin*dIII	5′-A↓AGCTT-3′ 3′-TTCGA↑A-5′	5′-A 3′-TTCGA AGCTT-3′ A-5′

DID YOU KNOW ?

Maps and Libraries

Restriction endonucleases are also used to create genetic maps and libraries. Go to the Nelson Web site for information on these applications.

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Figure 4 shows the action of the restriction enzyme *Eco*RI. *Eco*RI scans a DNA molecule and stops when it is able to bind to its recognition site. Once bound to the site, it cuts the bond between the guanine and adenine nucleotides on each strand. At the end of each cleavage site, one strand is longer than the other and has exposed nucleotides that lack complementary bases. The overhangs produced by the exposed DNA nucleotides are called **sticky ends**. *Eco*RI always cuts between the guanine and the adenine nucleotide on each strand. Since A and G are at opposite ends of the recognition site on each of the complementary strands, the result is the overhang, or sticky end, at each cleavage site.

sticky ends fragment ends of a DNA molecule with short single-stranded overhangs, resulting from cleavage by a restriction enzyme

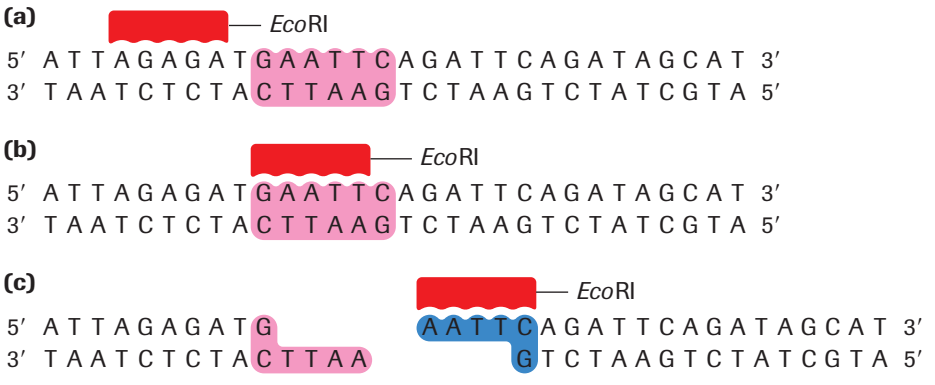

Figure 4
Cleavage of DNA sequence using restriction enzyme *Eco*RI.
(a) *Eco*RI scans the DNA molecule.
(b) *Eco*RI binds to the recognition site.
(c) *Eco*RI cuts between the guanine and adenine nucleotides, producing two fragments with complementary ends.

Not all restriction endonucleases produce sticky ends. For example, the restriction endonuclease *Sma*I produces **blunt ends**, which means that the ends of the DNA fragments are fully base paired (**Table 2**). Since *Sma*I cuts between the cytosine and guanine nucleotides and since these nucleotides are directly opposite each other in their complementary strands, the result is a blunt cut without sticky ends.

blunt ends fragment ends of a DNA molecule that are fully base paired, resulting from cleavage by a restriction enzyme

Restriction endonucleases that produce sticky ends are a generally more useful tool to molecular biologists than those that produce blunt ends. Sticky-end fragments can be joined more easily through complementary base pairing to other sticky-end fragments that were produced by the same restriction endonuclease. However, this is not always possible. To create recombinant DNA, molecular biologists choose restriction enzymes that will not cut in the middle of the DNA sequence of interest. For example, if the goal is to create recombinant DNA containing a particular gene, you would avoid using a restriction enzyme that cuts within the sequence of that gene.

Restriction enzymes are named according to the bacteria they come from. Generally speaking, the first letter is the initial of the genus name of the organism. The second and third letters are usually the initial letters of the species name. The fourth letter indicates the strain, while the numerals indicate the order of discovery of that particular enzyme from that strain of bacteria.

Learning Tip

Restriction enzymes are named according to specific rules. For example, the restriction enzyme *Bam*HI is named as follows:

- *B* represents the genus *Bacillus.*
- *am* represents the species *amyloliquefaciens.*
- *H* represents the strain.
- *I* means that it was the first endonuclease isolated from this strain.

Practice

1. The following sequence of DNA was digested with the restriction endonuclease *Sma*I:

   5′-AATTCGCCCGGGATATTACGGATTATGCATTATCCGCCCGGGATATTTTAGCA-3′

   3′-TTAAGCGGGCCCTATAATGCCTAATACGTAATAGGCGGGCCCTATAAAATCGT-5′

   *Sma*I recognizes the sequence CCCGGG and cuts between the C and the G.
   (a) Copy this sequence into your notebook and clearly identify the location of the cuts on it.
   (b) How many fragments will be produced if *Sma*I digests this sequence?
   (c) What type of ends does *Sma*I produce?
2. *Hin*dIII recognizes the sequence AAGCTT and cleaves between the two A's. What type of end is produced by cleavage with *Hin*dIII?
3. Explain why restriction endonucleases are considered to be molecular tools.
4. Copy the following sequence of DNA into your notebook. Write out the complementary strand. Clearly identify the palindromic sequences by circling them.

   GCGCTAAGGATAGCATTCGAATTCCCAATTAGGATCCTTTAAAGCTTATCC

DID YOU KNOW?

The First Restriction Enzyme

The first restriction endonuclease, *Hin*dIII, was identified in 1970 by Hamilton Smith at John Hopkins University. Smith received the Nobel Prize in 1978 for his discovery. Since then, more than 2500 restriction endonucleases have been identified.

Methylases

Methylases are enzymes that can modify a restriction enzyme recognition site by adding a methyl ($-CH_3$) group to one of the bases in the site (**Figure 5**). Methylases are important tools in recombinant DNA technology. They protect a gene fragment from being cut in an undesired location.

methylase an enzyme that adds a methyl group to one of the nucleotides found in a restriction endonuclease recognition site

Like restriction enzymes, methylases were first identified in bacterial cells. Methylases are used by a bacterium to protect its DNA from digestion by its own restriction enzymes. In bacteria, restriction enzymes provide a crude type of immune system. In fact, the term *restriction* comes from early observations that these enzymes appeared to restrict the infection of *E. coli* cells by viruses known as bacteriophages. The restriction enzymes bind to recognition sites in the viral DNA and cut it, making it useless. Eukaryotic cells also contain methylases. However, in eukaryotes methylation usually occurs in order to inactivate specific genes.

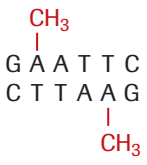

Figure 5
At a methylated *Eco*RI site, *Eco*RI restriction enzyme is no longer able to cut.

DID YOU KNOW ?

Eukaryotic Methylation

Methylases in eukaryotes are connected with the control of transcription. In addition, approximately 2 % of mammalian ribosomal RNA is methylated after it is transcribed.

DNA Ligase

To create recombinant DNA, pieces of DNA from two sources must be joined together. Using restriction enzymes and methylases, molecular geneticists can engineer fragments of DNA that contain the specific nucleotide sequences they want. These segments of DNA are then joined together by DNA ligase. If two fragments have been generated using the same restriction enzyme, they will be attracted to each other at their complementary sticky ends. Hydrogen bonds will form between the complementary base pairs. DNA ligase then joins the strands of DNA together (**Figure 6**).

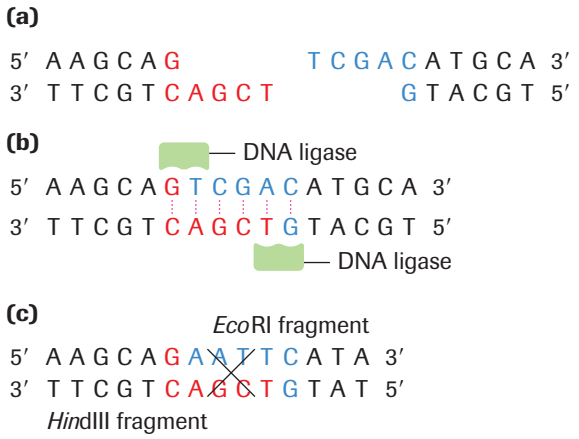

Figure 6

DNA ligase is able to join complementary sticky ends produced by the same restriction enzyme via a condensation reaction.

(a) Complementary sticky ends produced by *Hin*dIII

(b) Hydrogen bonds form between complementary bases. DNA ligase creates bonds between nucleotides in the DNA backbones.

(c) If fragments are not complementary, then hydrogen bonds will not form.

Taq DNA Polymerase and the Polymerase Chain Reaction

polymerase chain reaction (PCR) a technique for amplifying a DNA sequence by repeated cycles of strand separation and replication

In 1985, American scientist Kary Mullis invented a biotechnology technique called the **polymerase chain reaction (PCR)**. PCR allows scientists to make billions of copies of pieces of DNA from extremely small quantities of DNA. The reaction depends on the special property of *Taq* polymerase. In nature, *Taq* DNA polymerase is found in the bacterium *Thermos aquaticus*, which lives at extremely high temperatures. Like all the DNA polymerases, *Taq* DNA polymerase synthesizes DNA during replication. As you have learned previously, enzymes have an optimum temperature range in which they function. One adaptation that allows *Thermos aquaticus* to survive at high temperatures is that its DNA polymerase is stable at much higher temperatures than DNA polymerases from other organisms. Mullis used the heat-stable property of *Taq* polymerase in his PCR technique.

To prepare for PCR, the following materials are placed together in a small tube: *Taq* polymerase, the DNA to be copied, a large quantity of the four deoxynucleotides (A, T, G, and C), and short primers. The tube is then inserted into a PCR machine. PCR involves four simple steps (**Figure 7**).

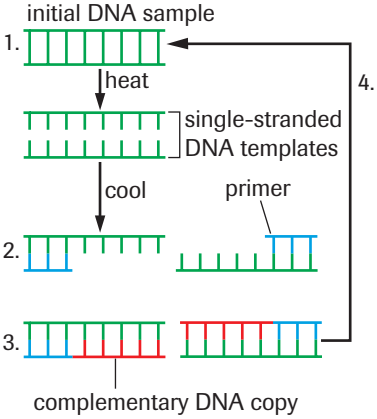

Figure 7

Steps in the PCR

1. The mixture is heated to a temperature high enough to break the hydrogen bonds in the double helix of the DNA and separate the strands. This forms single-stranded DNA templates.
2. The mixture is cooled, and the primers form hydrogen bonds with the DNA templates.
3. *Taq* polymerase synthesizes a new stand of DNA from the DNA template by complementary base pairing, starting at each primer.
4. The cycle of heating and cooling is repeated many times.

Each PCR cycle doubles the number of DNA molecules. After just 10 cycles there are 2^{10} (over two million) copies of the DNA template. Since scientists can use PCR to synthesize many identical copies from a very small sample of DNA, this technology has led to many advances in medicine, evolutionary biology, genetic engineering, and forensic science. Mullis was awarded the Nobel Prize in Chemistry in 1993 for his invention.

INVESTIGATION 20.2 ***Introduction***

Restriction Enzyme Digestion of Bacteriophage DNA

Using restriction enzymes and electrophoresis, molecular biologists are able to excise and isolate target sequences from DNA. How would the banding patterns compare if the same fragment of DNA were digested with different restriction enzymes? In this investigation, you will conduct electrophoresis of bacteriophage DNA that has been digested with restriction enzymes.

Report Checklist

● Purpose	● Design	● Analysis
○ Problem	○ Materials	● Evaluation
● Hypothesis	○ Procedure	○ Synthesis
● Prediction	● Evidence	

To perform this investigation, turn to page 696.

Transformation

So far, you have seen that mapping and sequencing can be used to identify the relative position and nucleotide sequence of genes in a DNA molecule. Using various enzymes, scientists can isolate DNA fragments containing a gene or genes. Multiple copies of the fragment can be prepared using PCR. The DNA fragment may also be joined (annealed) to other DNA fragments.

In genetics, transformation is any process by which foreign DNA is incorporated into the genome of a cell. A **vector** is the delivery system used to move the foreign DNA into a cell. The specific vector used for transformation is chosen based on the size and sequence of the foreign DNA fragment, the characteristics of the cells to be transformed, and the goal of the transformation. The goal of most genetic transformation is to express the gene product(s), and so change the traits of the organism that receives the foreign DNA. An organism with foreign DNA in its genome is said to be **transgenic**.

vector a vehicle by which foreign DNA may be introduced into a cell

transgenic a cell or an organism that is transformed by DNA from another species

plasmid a small double-stranded circular DNA molecule found in some bacteria

Transformation of Bacteria

Bacteria are the most common organisms that are transformed by molecular biologists. Transgenic bacteria may be used to study gene expression or gene function, to create and maintain a stock of a particular DNA fragment, or to synthesize a useful gene product. For example, transgenic bacteria have been engineered to produce human growth hormone, used in the treatment of pituitary dwarfism.

The first stage of transformation for any organism is to identify and isolate the DNA fragment that is to be transferred. The DNA fragment is then introduced into the vector. **Plasmids** are small, circular, double-stranded DNA molecules that occur naturally in the cytoplasm of many bacteria (**Figure 8**). Plasmids are commonly used as vectors for bacterial transformation. A plasmid contains genes, and it is replicated and expressed independently of the large bacterial chromosome. There can be many copies of a plasmid in a single bacteria cell and, under certain conditions, plasmids can pass through the cell membrane.

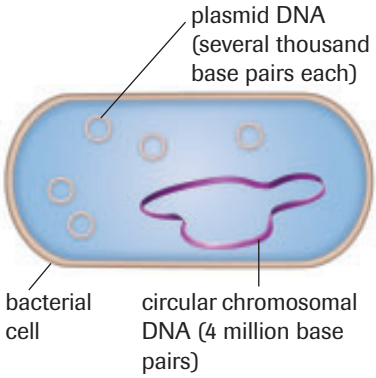

Figure 8
Chromosomal and plasmid DNA coexist in many bacteria.

Figure 9, on the next page, is a diagram of the basic steps in producing transgenic bacteria. First, both the plasmid vector and the DNA containing the desired sequence are cut by the same restriction enzyme(s). In this example, both DNA molecules are cut by *Eco*RI, generating sticky ends. The cut plasmid and DNA fragment are then mixed together and incubated with DNA ligase. This produces recombinant plasmids that

EXTENSION

How to Make cDNA

One way to make copies of a particular gene is to use an enzyme called reverse transcriptase. This enzyme synthesizes DNA from mRNA. The resulting molecule is called copy DNA or cDNA. The cDNA can then be transferred into a vector or a cell.

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contain the foreign DNA fragment. The bacterial cells are then treated to open pores in the cell membrane, which allows them to take up the recombinant plasmid. Once a bacterium has been transformed, it makes many copies of the recombinant plasmid, each of which includes a copy of the foreign DNA. This is often called gene cloning since the bacterium produces many identical copies (clones) of the original DNA fragment.

However, not all the bacterial cells will take up the recombinant plasmid. How can a scientist or technician distinguish between bacteria with a plasmid and those without? Plasmids used for transformation experiments often carry genes for antibiotic resistance, which can then be used to select for transformed bacteria. By growing the bacteria in medium that contains the antibiotic, any cells that do not contain a plasmid are killed off. Individual bacteria cells are then grown into colonies so that the plasmid DNA can be isolated from the cells and checked to make sure it contains the desired foreign DNA sequences.

For this transformation procedure to be successful, the plasmid DNA must have only one recognition site for the restriction enzyme that is used, or else it would be cut into a number of useless pieces. Naturally occurring plasmids do not always have a single appropriate restriction enzyme site, so scientists have engineered plasmids especially for transformation. Most of these engineered plasmids contain a **multiple-cloning site**, which is a single region that contains unique recognition sites for an assortment of restriction enzymes. The recognition sites are positioned very close together and are not found anywhere else on the plasmid's DNA sequence.

multiple-cloning site a region in a vector that is engineered to contain the recognition site of a number of restriction enzymes

Vectors other than plasmids may also be used to transform bacteria, including viruses and small inert particles that are literally fired into the cells.

DID YOU KNOW?

Plasmids: Beneficial Guests

Japanese scientists were the first to discover plasmids that carry genes for multiple drug resistance. The bacterium *Shigella*, which causes dysentery, developed resistance to as many as four antibiotics, including tetracycline, streptomycin, chloramphenicol, and the sulfonamides. The multidrug resistance was due to a plasmid within the bacterium that carried genes for resistance and could be passed naturally from bacterium to bacterium.

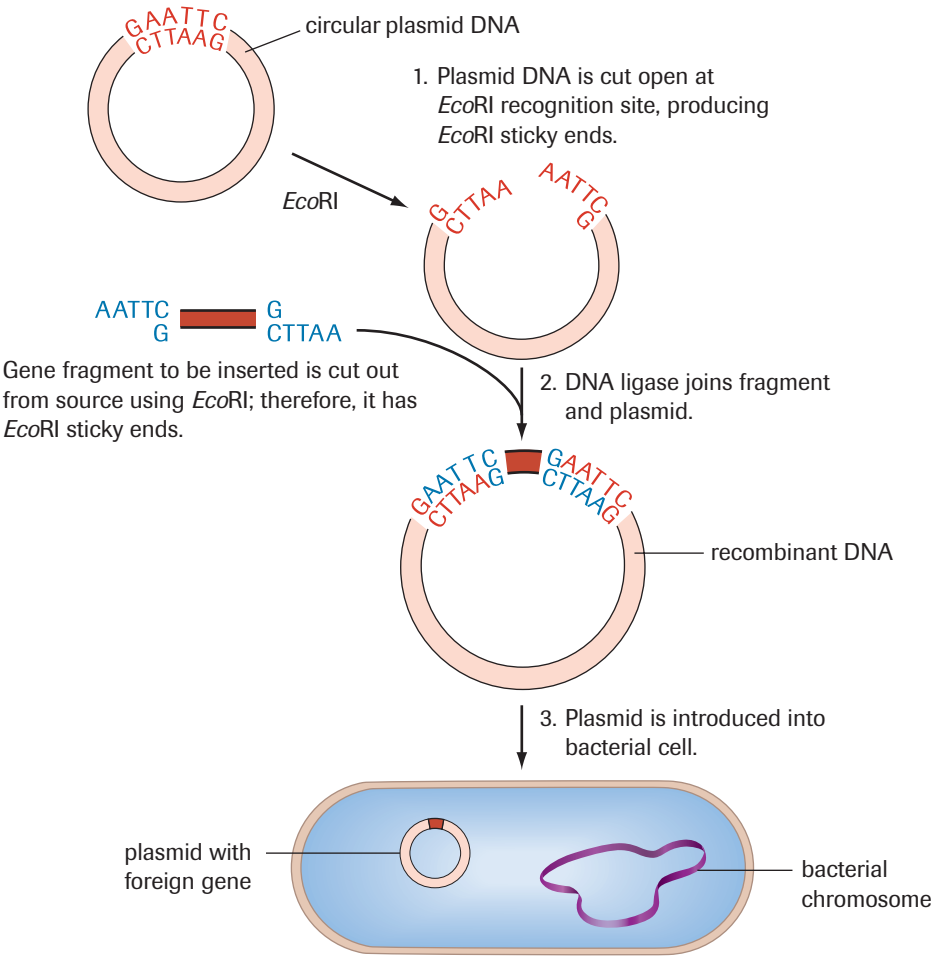

Figure 9
A foreign gene is introduced into a plasmid. The plasmid is now an example of recombinant DNA, which can be introduced into a bacterial cell to produce numerous copies of the gene.

WEB Activity

Case Study—Transformation of Eukaryotes

The first transgenic animal and the first transgenic plant were both produced in 1982. The animal was a mouse that contained the gene for growth hormone from a rat. The plant was a tobacco plant that contained a gene from a bacterial cell. The introduced gene produced an antibiotic in the plant's cells that protected the plant from bacterial infection. Since then, many transgenic animals and plants have been produced.

Producing transgenic eukaryotes is a lot more complex than the transformation of bacteria, and new techniques are still being developed. In this activity, you will find out about one technique used to create transgenic eukaryotes.

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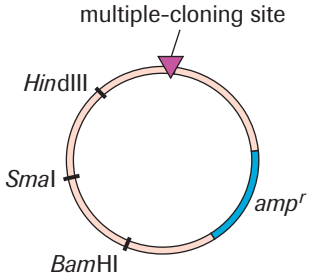
SUMMARY *DNA and Biotechnology*

Table 3 Key Tools of Molecular Biology

Tool	Use	Example
restriction endonuclease	bacterial enzyme that cuts DNA sequences at a specific recognition site	*Bam*HI recognition site: 5′-GGATCC-3′ 3′-CCTAGG-5′ DNA sequence before cleavage: 5′-TCAGCGGATCCCAT-3′ 3′-AGTCGCCTAGGGTA-5′ DNA sequence after cleavage with *Bam*HI: 5′-TCAGCG GATCCCAT-3′ 3′-AGTCGCCTAG GGTA-5′
methylase	enzyme that adds a methyl group to recognition sites to protect DNA from cleavage by restriction enzyme	*Bam*HI methylase adds methyl group ($-CH_3$) to second guanine nucleotide in the recognition site: 5′-GGATCC-3′ 3′-CCTAGG-5′ DNA sequence no longer cleaved by *Bam*HI methyl group changes recognition site
DNA ligase	enzyme that joins DNA fragments by creating bonds between nucleotides in the DNA backbone	DNA fragments before subjection to DNA ligase: 5′-ATAGTG-3′ 5′-AATTCGG-3′ 3′-TATCACTTAA-5′ 3′-GCC-5′ DNA fragments after subjection to DNA ligase: 5′-ATAGTGAATTCGG-3′ 3′-TATCACTTAAGCC-5′ two fragments are joined
plasmid	small circular DNA that has the ability to enter and replicate in bacterial cells and, therefore, can be used as a vector to introduce new genes into a bacterial cell	plasmid containing multiple-cloning site, ampicillin-resistance gene, and other restriction enzyme sites 

Section 20.3 Questions

1. Define *restriction endonuclease* and *methylase*.
2. Restriction endonucleases are found in many species of bacteria.
   (a) Describe their role and function in a bacterial cell.
   (b) How does the role of restriction endonucleases in nature differ from the role of restriction endonucleases in the laboratory setting?
3. Distinguish between blunt ends and sticky ends.
4. Define *recognition site*. Using examples to support your answer, depict the palindromic nature of recognition sites.
5. Restriction enzymes cut at recognition sites that are usually six to eight base pairs in length. Provide reasons why a 2-base-pair recognition site would be too short to be useful and a 14-base-pair recognition site may be too long to be useful in the field of genetic engineering.
6. Sketch a diagram that summarizes the process of polymerase chain reaction (PCR). Clearly label the important features.
7. Explain why the Human Genome Project's initial years were spent developing techniques that would sequence larger DNA strands efficiently. (*Hint:* The human genome contains approximately three billion base pairs.)
8. As a scientist working for a pharmaceutical company, you are asked to engineer bacteria that will produce human growth hormone. The objective is commercial production in order to treat individuals who are deficient in this hormone. Describe the steps you would take in order to produce this hormone.
9. Transformation technology is used in agriculture to create genetically modified organisms (GMOs) that contain useful traits. This is a controversial technology, however. Some people think that GMOs pose unacceptable environmental or health risks. The Government of Canada has set regulations that must be met for approval of GMOs. Using the Internet and other resources, research the regulations that have been put into place. Do you feel these guidelines are adequate? What modification would you make to these guidelines if you could? Explain the implications of the guidelines that have been set.

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Extension

10. In order to create recombinant DNA containing the desired sequences, scientists have developed a number of procedures to find and isolate DNA, and to confirm whether a transgenic organism contains the foreign DNA. Go to the Nelson Web site to find out how the techniques of electrophoresis, Southern blotting, and Northern blotting work and when they are used. Then, summarize the information in a chart or another appropriate format.

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11. PCR can be used to create a DNA "fingerprint" that can identify an individual. This technique has been applied to forensics. In some well-known cases, such as that of Guy Paul Morin, PCR has been used to overturn convictions made before the technology was available. In June 2000, the Government of Canada passed the DNA Identification Act, which gave the Royal Canadian Mounted Police the right to create and maintain a database of DNA fingerprints. Conduct research on the use of PCR to identify individuals. Then, use this information to prepare a convincing argument for or against the requirement that anyone accused of a serious crime must supply police with a DNA sample.

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Mutations and Genetic Variation 20.4

Mutations are changes in the sequence of the DNA molecule and are the source of new genetic variation that may be acted on by natural selection. A beneficial mutation gives an organism a selective advantage and tends to become more common over time, leading to new evolutionary changes. A harmful mutation reduces an individual's fitness and tends to be selected against. Harmful mutations occur at low rates in a species. Some mutations are neutral, having neither a benefit nor a cost, and are not acted on by natural selection.

As scientists gained more knowledge about the nature of DNA and the genetic code, they were able to more fully understand mutations. **Point mutations** are changes in a single base pair of a DNA sequence. They may or may not change the sequence of amino acids. **Gene mutations** change the amino acids specified by the DNA sequence, and they often involve more than a single base pair. **Figure 1** summarizes the DNA changes that occur in some common types of mutation.

Learning Tip

Review how mutations contribute to the variability of species and how natural selection acts on mutations in Chapter 6 of this book.

point mutation a mutation at a specific base pair

gene mutation a mutation that changes the coding for amino acids

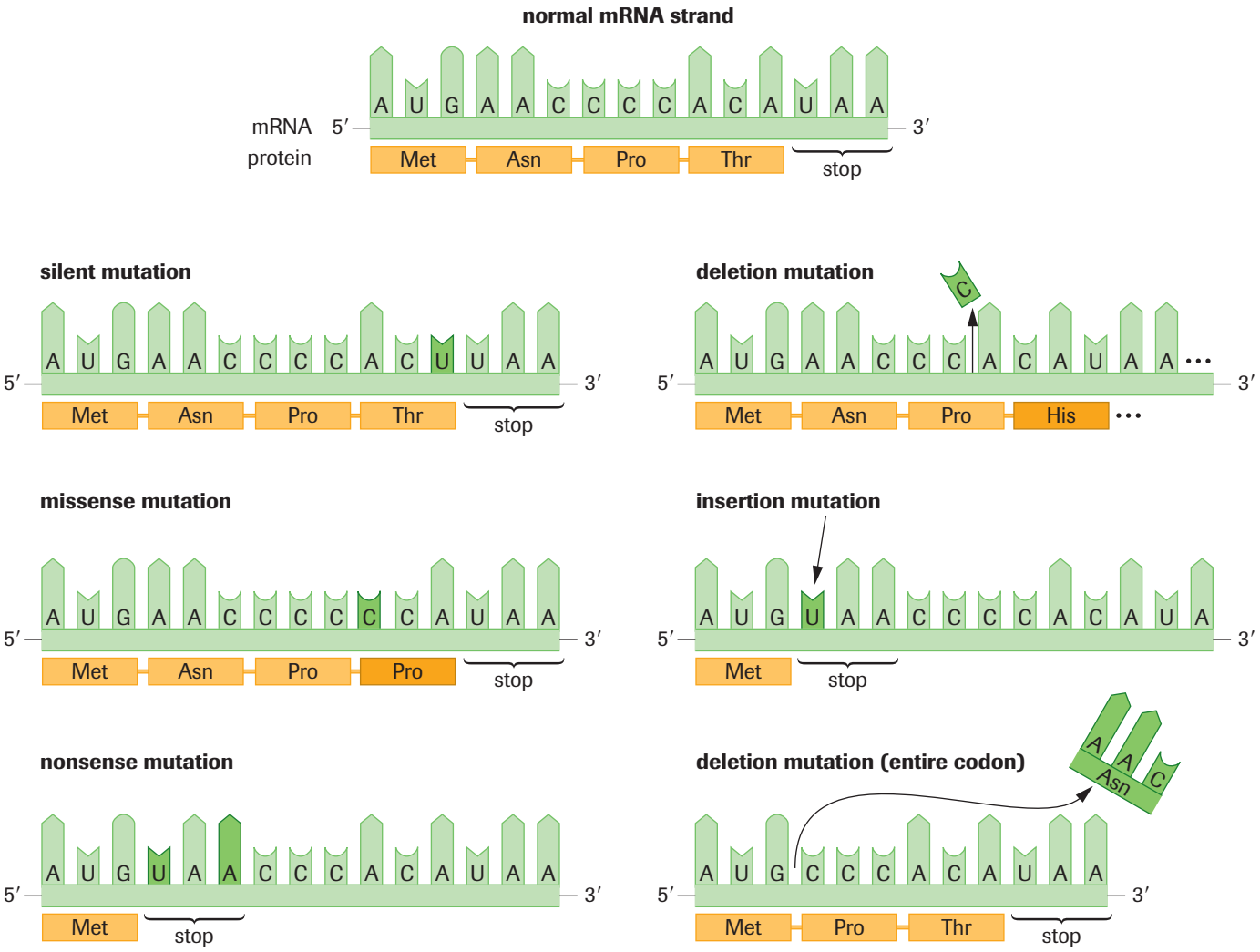

Figure 1
A summary of different types of mutations that may occur in a DNA sequence, affecting the transcribed RNA sequence.

silent mutation a mutation that does not result in a change in the amino acid coded for

One type of point mutation, called a **silent mutation**, has no effect on the operation of the cell. In the silent mutation example in **Figure 1**, the codon for threonine has changed from ACA to ACU. However, this mutation does not change the amino acid because both these codons code for threonine. Most silent mutations occur in the non-coding regions, so they do not affect protein structure.

missense mutation a mutation that results in the single substitution of one amino acid in the polypeptide

A **missense mutation** arises when a change in the base sequence of DNA alters a codon, leading to a different amino acid being placed in the polypeptide. Sickle cell anemia is the result of a missense mutation. Another type of point mutation is a nonsense mutation. A **nonsense mutation** occurs when a change in the DNA sequence causes a stop codon to replace a codon specifying an amino acid. During translation, only the part of the protein that precedes the stop codon is produced, and the fragment may be digested by cell proteases. Nonsense mutations are often lethal to the cell. Missense and nonsense mutations arise from the substitution of one base pair for another.

nonsense mutation a mutation that converts a codon for an amino acid into a stop codon

deletion the elimination of a base pair or group of base pairs from a DNA sequence

An example of a gene mutation is a **deletion**, which occurs when one or more nucleotides are removed from the DNA sequence. In the deletion mutation example in **Figure 1**, on the previous page, a cytosine nucleotide has been deleted. This changes the third codon from CCC to CCA, but the amino acid does not change because both CCC and CCA code for proline. However, the deletion also causes a change in the fourth codon, from ACA to CAU. This does affect the amino acid, changing it from threonine to histidine. Such shifts in the reading frame usually result in significant changes to the protein.

insertion the placement of an extra nucleotide in a DNA sequence

Another way that a shift in the reading frame can occur is by the **insertion** of a nucleotide. Since the DNA sequence is read in triplets of nucleotides, inserting an extra nucleotide will cause different amino acids to be translated, similar to a deletion mutation. When a mutation changes the reading frame, it is called a **frameshift mutation**. Insertions and deletions can both cause frameshift mutations. A deletion or insertion of two nucleotides will also result in a shift of the reading frame; however, a deletion or insertion of three nucleotides does not have this effect. Instead, the insertion or deletion of three nucleotides results in the addition or removal of one amino acid.

frameshift mutation a mutation that causes the reading frame of codons to change

translocation the transfer of a fragment of DNA from one site in the genome to another location

Another category of mutations involves large segments of DNA and is seen at the chromosomal level. **Translocation** is the relocation of groups of base pairs from one part of the genome to another. Usually translocations occur between two nonhomologous chromosomes. A segment of one chromosome breaks and releases a fragment, while the same event takes place on another chromosome. The two fragments exchange places, sometimes disrupting the normal structure of genes. When unrelated gene sequences come together and are transcribed and translated, the result is a fusion protein with a completely altered function, if any. Some types of leukemia are associated with translocations and their respective fusion proteins.

inversion the reversal of a segment of DNA within a chromosome

Finally, an **inversion** is a section of a chrosome that has reversed its orientation in the chromosome (has turned itself around). There is no gain or loss of genetic material, but, depending on where the inversion occurs, a gene may be disrupted.

Causes of Genetic Mutations

spontaneous mutation a mutation occurring as a result of errors made in DNA replication

Some mutations are simply caused by error of the genetic machinery and are known as **spontaneous mutations**. For example, DNA polymerase I occasionally misses a base or two, which results in a point mutation. Mutations may also arise from exposure to **mutagenic agents**. These are **induced mutations**. Some examples of mutagenic agents include ultraviolet (UV) radiation, cosmic rays, X-rays, and certain chemicals.

mutagenic agent an agent that can cause a mutation

induced mutation a mutation caused by a chemical agent or radiation

Case Study

Gene Mutations and Cancer

Cancer is considered a genetic disease because it is always associated with a mutation in the genetic sequence. However, many different things can alter DNA, including viruses and various environmental factors (**Figure 2**).

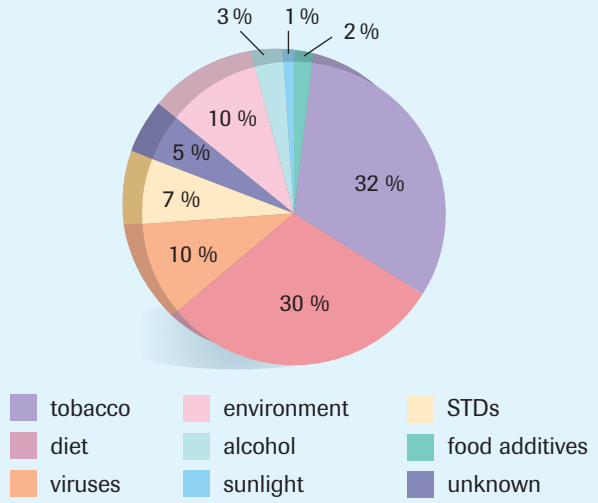

Figure 2
Estimates of risk factors for cancer calculated in percentages. Lifestyle choices related to diet and smoking can be linked with over 60 % of cancer cases.

Viruses inject foreign genetic information into cells, disrupting the DNA that codes for cell division. Some viruses that are linked to sexually transmitted diseases are known to cause cancer. For example, women who have human papillomavirus (HPV) have a greater incidence of cancer. Environmental factors have been linked to other types of cancer. Skin cancer, for example, has been linked with ultraviolet radiation from the Sun. Exposure to harmful chemicals in our environment can also cause cancer. A number of cancer-causing substances can be found in cigarettes.

Whatever the initial cause, scientists agree that all cancers are related to mutations. Usually, it takes more than one mutation to trigger a malignant growth. This is why cancer usually occurs more frequently in older people.

Two lines of evidence indicate that cancer results from mutations. First, cancer cells often display nitrogen base substitution, or the movement of genetic material from one part of the chromosome to another. Second, many known mutagens are also known to cause cancer. X-rays, ultraviolet radiation, and mutagenic chemicals can induce cancer.

In 1982, molecular biologists were able to provide additional evidence to support the hypothesis that cancer could be traced to genetic mutations. Segments of chromosomes extracted from cancerous mice were used to transform normal mouse cells (growing in tissue culture) into cancerous cells. The cancer-causing genes, called oncogenes, seemed to turn on cell division. In their noncancerous state, oncogenes are usually referred to as proto-oncogenes. Proto-oncogenes may remain inactive or may perform some useful function until they are triggered to become active oncogenes. Evidence suggests that activation occurs in a number of steps, so a single “hit” (mutation) does not immediately result in cancerous cell divisions.

Further studies indicate that cancer-causing oncogenes are present in normal strands of DNA. But if oncogenes are found in normal cells, why do normal cells not become cancerous? One current theory that has gained acceptance from the scientific community suggests that the cancer gene has been transposed (moved) to another gene site. Such transpositions may have been brought about by environmental factors or mutagenic chemicals or other agents.

Genes that direct the assembly of amino acids into proteins are referred to as structural genes. Genes called regulator genes act like a switch to turn “off” segments of the DNA molecule, so that a gene is active only when and where its gene product is needed. In very simple terms, when a mutagen causes the oncogene to become separated from its regulator gene, the cell may then be unable to turn the gene "off" (**Figure 3**). This causes the cell to continue to divide at an accelerated rate.

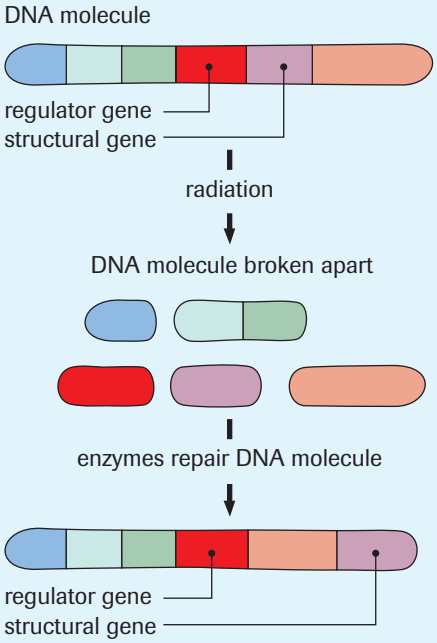

Figure 3
Mutagenic agents may cause the separation of the regulator and structural genes. If the structural gene codes for a protein involved in controlling cell division, this separation can lead to cancer.

The most common oncogene, *ras*, is found in 50 % of colon cancers and 30 % of lung cancers. Present in normal cells, *ras* makes a protein that acts as an "on" switch for cell division. *Ras* ensures that cells divide to replace damaged or dead cells. After a sufficient number of cells have been produced, the *ras* gene should be turned off. But the cancer-causing oncogene produces a protein that blocks the "off" switch. With the switch left on, cell division goes on continuously.

Case Study Questions

1. Why do many scientists believe that certain viruses cause cancer?
2. How does sunlight cause cancer?
3. List three environmental carcinogens and suggest a possible source for each.
4. Distinguish between oncogenes and proto-oncogenes.
5. Explain how oncogenes are activated.
6. What is the *ras* gene?

Inferring Relationships from DNA Sequences

At one time, scientists could compare and classify species based only on their morphology and behaviour. For example, Charles Darwin found evidence for the theory of evolution by comparing anatomical features of different species (see Chapter 6). Today, biologists can compare the genetic makeup of different species for evidence of relationships among them.

phylogeny proposed evolutionary history of a species or group of organisms

Phylogeny is the proposed evolutionary history of a group of organisms, or of a species. Overall, species that are closely related will share very similar DNA sequences, while those that are more distantly related will have more genetic differences. For example, you might expect that the sequence of DNA in a house cat's genome would have more similarities to that of a lion than to a sparrow. As we have seen, the DNA of any organism can mutate. Natural selection acts on beneficial and harmful mutations in a population, changing the relative proportions of these mutations that are passed on from generation to generation. The genomes of two species with a recent common ancestor would have had less time and opportunity for mutations to accumulate and be selected, and so we can predict that they would show fewer differences.

Learning Tip

Lab Exercise 5.A in Chapter 5 shows an example of how differences in genomic DNA sequences provide evidence for the relationships among various species.

Mutations do not occur only in genomic DNA. Nuclear DNA is often quite a large genome, so for some research it is more efficient for scientists to examine the changes in the smaller genomes of mitochondria or chloroplasts. In particular, mitochondrial DNA (mtDNA) can be used to trace inheritance through the maternal line in mammals, as the egg is the only source of the mitochondria that are passed on to new offspring.

Mitochondrial DNA has also provided some fascinating clues about the evolutionary history of modern humans. Two theories are proposed to explain the current distribution of humans around the world. One proposes that modern humans, *Homo sapiens*, evolved simultaneously in different regions of the world from an earlier species, *Homo erectus*. This theory is called the multiregional model and proposes that the different ethnic groups observed worldwide today would have begun their evolution to *Homo sapiens* between one and two million years ago. According to this model, the groups interbred to some degree, and so didn't form into different species. The second theory, called the monogenesis model, proposes that *Homo* species moved out of Africa twice: first as *Homo erectus*, and second as *Homo sapiens* between 100 000 and 200 000 years ago, and that modern ethnic groups are all descendants of the second migration.

DID YOU KNOW?

The Romanovs

Mitochondrial DNA was used to identify the suspected remains of the imperial Romanov family in Russia, who were murdered by the Bolsheviks in 1918. To do so, mitochondrial DNA from Prince Philip of England, a close relative of the former Tsarina Alexandra through his maternal side, was compared to mitochondrial DNA recovered from the remains, resulting in positive identification and the resolution of an 80-year-old mystery.

Mitochondrial DNA analyses for a variety of individuals, representing the ethnic groups found around the world, seem to support the monogenesis model. The greatest variety of mtDNA mutations exist in African ethnic groups, which is consistent with the theory that mutations accumulate over time and that the population that has existed the longest will demonstrate the largest accumulation of mutations. Additionally, the mtDNA from ethnic groups on continents other than Africa were traced back to Africa rather than to each other.

Interspersed Elements

Other DNA analyses focus on intervening sequences inserted into DNA. For example, **SINEs** (**s**hort **in**terspersed **e**lements) and **LINEs** (**l**ong **in**terspersed **e**lements) are often associated with the genes of retroviruses within the genome and are thought to have been inserted by those viruses. SINEs and LINEs are often located in areas of the DNA that appear to be noncoding regions. That is, the DNA in these areas does not code for one of the known gene products of that species. Although the function of the DNA in these regions is not known, it is inherited; therefore, changes to these DNA sequences, such as insertions, are passed to succeeding generations.

If two species have the same SINE or LINE located at precisely the same position in their DNA, it can be assumed that the insertion occurred only once in a common ancestor. SINEs and LINEs make ideal markers for tracing evolutionary pathways. They are easy to find and identify, even if they undergo small mutational changes, because they are relatively large and recognizable segments of DNA often hundreds of base pairs in length. The possibility of a mutation reverting to an older form is extremely remote, as the chances of a SINE or LINE being inserted in exactly the same location in two different species is highly unlikely.

SINEs repeated DNA sequences 300 base pairs long that alternate with lengths of DNA sequences found in the genomes of higher organisms

LINEs repeated DNA sequences 5000 to 7000 base pairs long that alternate with lengths of DNA sequences found in the genomes of higher organisms

LAB EXERCISE 20.B

Looking for SINEs of Evolution

Report Checklist

○ Purpose	○ Design	● Analysis
○ Problem	○ Materials	● Evaluation
○ Hypothesis	○ Procedure	● Synthesis
○ Prediction	○ Evidence	

In this activity, you will use DNA sequences to predict and chart phylogenetic relationships among species.

Suppose you find a pattern in the noncoding SINE DNA of two different species, and do not find that pattern in other species. Evolution can explain the situation by saying that the two species recently had a common ancestor, and that both species inherited this pattern from their ancestor. The predicted family tree is shown in **Figure 4**.

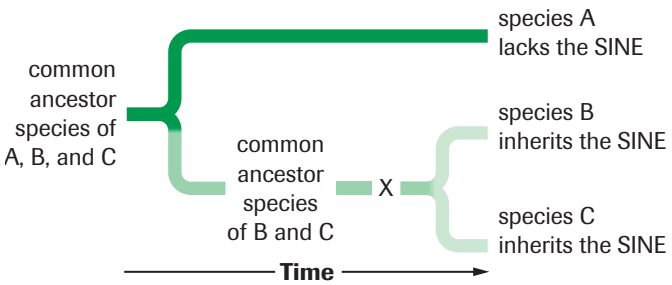

Figure 4
X indicates the time when the SINE became inserted into the genome. Since the SINE insertion occurs only once, at time X, the size and precise location of the SINE will be identical in species B and C.

Part I: Looking for a SINE

Procedure I

1. Examine the hypothetical DNA code from four different species (**Figure 5**). These species have large sections of DNA that appear to be homologous. These homologous sequences have been aligned vertically so that similarities and differences can be easily seen and colours are used to highlight those nucleotides that are not matches (**Figure 6**).

```
Species W   AGATAGCGCGTAAAAAG
Species X   AAATAGCGCGTAAATAG
Species Y   AAATAGTTAAAGTTACGCATAAATAC
Species Z   AGATAGCGCGTAAATGG
```

Figure 5
Sequenced DNA fragments from four distantly related species

```
Species W-   AGATAG.........CGCGTAAAAAG
Species X-   AAATAG.........CGCGTAAATAG
Species Y-   AAATAGTTAAAGTTACGCATAAATAC
Species Z-   AGATAG.........CGCGTAAATGG
```

Figure 6
DNA sequences from **Figure 5** aligned for comparison. Note that spaces appear in the sequences only to facilitate comparisons.

The single nucleotide differences have most likely resulted from point mutations, while the nine-nucleotide segment in species Y is probably the result of an insertion. (Note that this is much more likely than the alternative possibility—that each of the other species experienced an identical deletion event in its past.) The type of pattern observed in species Y often results from a SINE or LINE insertion.

2. Copy the DNA sequences in **Figure 7** into your notebook. Align the homologous sections vertically.
3. Use a highlighter to colour all positions that have the same nucleotide in all four species.
4. Use a different colour to highlight the SINE insertion.

Analysis and Evaluation I

(a) Identify any nucleotide differences in the SINE sequences. Explain how these differences might have occurred.

(b) Identify whether mutations that occur within the SINE are likely to be harmful, beneficial, or neutral. Explain.

(c) Based on the data alone, construct a chart similar to **Table 1** showing the phylogenetic relationship of these species.

Part II: Evolution Displayed by SINEs and LINEs

Procedure II

5. Study the data in **Table 1**. DNA sequencing was used to document the presence or absence of interspersed elements A through I in five mammals. Camels are included as the outgroup.

Table 1 Molecular Evidence for the Evolution of Whales*

Group	SINE or LINE								
	A	B	C	D	E	F	G	H	I
cow	+	+	−	−	−	−	+	−	+
pig	−	−	−	+	+	−	−	−	+
whale	−	+	+	−	−	−	+	+	+
deer	+	+	−	−	−	−	+	−	+
hippopotamus	−	+	+	−	−	+	+	+	+
camel	−	−	−	−	−	−	−	−	−

\+ indicates presence of element - indicates absence of element
* Data modified from Nikaido 1999

6. Use the data to construct a chart showing the phylogenetic relationships between these mammals. Clearly indicate the relative positions at which each insertion most likely occurred.

Analysis and Evaluation II

(d) Are whales more closely related to cows or hippopotamuses? Explain your rationale.

(e) Identify which insertion happened first: A or B? Explain your reasoning.

Synthesis

(f) Explain whether pigs and camels are more closely related than hippopotamuses and camels.

(g) What must be true about the genomes of all whale species (i.e., which SINEs must they all contain)? Explain your rationale.

(h) A researcher interested in the evolution of whales wants to know whether orcas are more closely related to white-sided dolphins or to pilot whales. Describe a way to answer this question.

```
Species P  ...AAATTGCTTCGTATTTTCGAATTGCCCCGCTAAAGCGCTTTAGC.......
Species Q  ...AACTTGCTTCGTATTAAGCTGTTGCGTAAAGTTAGTACGAATTGCCCCGGTGAAGCGCTTTAGC......
Species R  ...AATTTGCTTCGTTTTTTCGAATTGCCCCGCTAAAGCGCTTTAGC.......
Species S  ...AACTTGCTACGTATTAAGCCGTTGCGTAAAGTTAGGACGAATCGCCACGGTGACGCGCTTGAGC......
```

Figure 7
Homologous DNA sequences from four species

SUMMARY — Mutations and Genetic Variation

Table 2 Types of Mutations

Category	Type	Result
point mutation	**substitution** AAG CCC GGC AAA AAG ACC GGC AAA	**missense mutation** only one amino acid substituted
	deletion AAG CCC GGC AAA AAC CCG GCA AA ↑	**frameshift mutation** can result in many different amino acids substituted or a stop codon read (nonsense mutation)
	insertion AAG CCC GGC AAA AAG ACC GGG CAA A	
chromosomal	**translocation** chromosome 1 5′ AAATTCG GCACCA 3′ chromosome 2 5′ TAGCCC AAGCGAG 3′ ↓ chromosome 1 5′ TAGCCC GCACCA 3′ chromosome 2 5′ AAATTCG AGCGAG 3′	inactivation of gene if translocation or inversion is within a coding segment
	inversion normal chromosome 5′ AATTGGCCATA ATATGAA AAGCCC 3′ 3′ TTAACCGGTAT TATACTT TTCGGG 5′ ↓ after inversion 5′ AATTGGCCATA TTCATAT AAGCCC 3′ 3′ TTAACCGGTAT AAGTATA TTCGGG 5′	

- In mammals, mitochondrial DNA can be used to trace inheritance through the maternal lineage.
- Comparisons of DNA sequences can provide detailed phylogenetic relationships by revealing the specific changes in the genetic makeup of species and populations.
- SINEs and LINEs provide excellent inheritable markers for tracing the evolution of species' lineages.

Section 20.4 Questions

1. Clearly define the following terms and give an example of each: *mutation, frameshift mutation, point mutation, nonsense mutation, missense mutation*.
2. Explain why mutations, such as insertions or deletions, are often much more harmful than nitrogen-base substitutions.
3. Which of two types of mutations, nonsense or missense, would be more harmful to an organism? Explain your answer using your knowledge of protein synthesis.
4. Identify three factors that can produce gene mutations.
5. Identify the type of mutation that has occurred in the strands below. Describe the effect on the protein. The original strand is

   AUG UUU UUG CCU UAU CAU CGU

   Determine whether or not the following mutations would be harmful to an organism. Translate the mRNA sequence into protein to help you decide. The mutation is indicated in red.

   (a) AUG UUU UUG CCU UAU CAU CGU
   AUG UUU UUG CCU UAC CAU CGU

   (b) AUG UUU UUG CCU UAU CAU CGU
   AUG UUU UUG CCU UAA CAU CGU

   (c) AUG UUU UUG CCU UAU CAU CGU
   AUG UUU CUU GCC UUA UCA UCG U

   (d) AUG UUU UUG CCU UAU CAU CGU
   AUG UUU UUG CCU AUC AUC GU

   (e) AUG UUU UUG CCU UAU CAU CGU
   UGC UAC UAU UCC GUU UUU GUA
6. Which of the following amino acid changes can result from a single base-pair substitution?
   (a) arg to leu
   (b) cys to glu
   (c) ser to thr
   (d) ile to ser
7. Explain why a food dye that has been identified as a chemical mutagen poses greater dangers for a developing fetus than for an adult.
8. List three changes that can be made to your personal lifestyle that would reduce the odds of a mutation taking place.
9. Explain how mutations may be of benefit to an organism, and describe how these beneficial mutations are maintained in a species. Identify the biological process that influences which mutations stay in a population over time.
10. Both mitochondria and chloroplasts contain their own genomes, which are separate from the nuclear genome. The DNA in mitochondria and chloroplasts have been used as evidence for the endosymbiotic theory of the evolution of eukaryotic organisms. This theory was developed by the American scientist Dr. Lynn Margulis. According to this theory, mitochondria and chloroplast arose from bacteria and algae cells that became engulfed by another cell with which they had a symbiotic relationship. Over time, the bacteria and algae became a part of the other cell. Evidence of this theory can be found by comparing the DNA of mitochondria with bacteria, and of chloroplasts with algae. Go to the Nelson Web site to learn more about the theory of endosymbiosis, and summarize the DNA evidence that supports it.

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Extension

11. The mutation that causes sickle cell anemia involves the substitution of the amino acid valine for the amino acid glutamic acid. Research the structure of valine and glutamic acid and, with your knowledge of chemistry, hypothesize why this substitution results in a large conformational change for the hemoglobin protein. List other amino acids that could have been substituted instead of valine that may not have caused such serious side effects. List amino acids that are similar to glutamic acid that would probably cause similar side effects.

INVESTIGATION 20.1

Protein Synthesis and Inactivation of Antibiotics

Report Checklist

● Purpose	● Design	● Analysis
○ Problem	○ Materials	● Evaluation
● Hypothesis	○ Procedure	● Synthesis
● Prediction	● Evidence	

In this investigation, you will examine the effects of ampicillin on two types of bacteria. *E. coli* MM294/pAmp contains a gene insert that directs the synthesis of a protein that inactivates ampicillin, whereas *E. coli* MM294 does not. Ampicillin inhibits bacterial growth by interfering with cell wall biosynthesis. Based on your knowledge of protein synthesis, make a prediction about the survival of *E. coli* MM294/pAmp and *E. coli* MM294 on ampicillin-rich media.

Problem

What effect does the presence of an ampicillin-resistance gene in a bacterium have on its growth on ampicillin-rich media?

Materials

apron	masking tape
safety goggles	permanent marker
gloves	inoculating loop
10 % bleach	Bunsen burner
2 LB agar plates	MM294 culture
2 LB + ampicillin (LB/amp) plates	MM294/pAMP culture
	37 °C incubator

Wear safety goggles at all times.

Wear gloves when performing the experiment. Disposable latex gloves are best avoided since allergic reactions to latex have been widely reported. Disposable polyethylene, PVC, or neoprene gloves are recommended.

Wipe down all surfaces with 10 % bleach before and after the laboratory exercise.

All resulting cultures must be immersed in 10 % bleach before disposal to ensure sterilization.

Do not leave a lit Bunsen burner unattended. Refer to Appendix C2 for a review of the safe use of a Bunsen burner.

Wash your hands thoroughly at the end of the laboratory.

Procedure

1. Put on your safety goggles and gloves, and wipe down your bench with a 10 % bleach solution.
2. Obtain two LB plates and two LB/amp plates from your teacher.
3. Label the bottom of each plate with your name and the date, using a permanent marker.
4. Label both of the LB plates "− amp" for the *E. coli* MM294 cells. Label both of the LB/amp plates "+ amp" for the *E. coli* MM294/pAMP cells.
5. Hold your inoculating loop like a pencil and sterilize it in the nonluminous flame of the Bunsen burner until it becomes red hot. Cool the sterilized loop by touching it to the edge of the agar on one of the LB plates.
6. Using the sterilized loop, pick up one colony of *E. coli* MM294 from a start culture plate. Glide the inoculating loop across an LB agar plate, making sure not to gouge the agar (**Figure 1**).

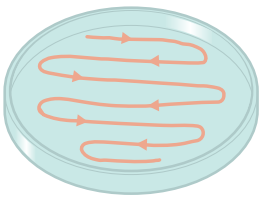

Figure 1
Pattern of streaking on an agar plate

7. Resterilize your loop as directed in step 5.
8. Repeat step 6 with *E. coli* MM294 streaked on an LB/amp plate.
9. Resterilize your loop as directed in step 5.
10. Repeat step 6 with *E. coli* MM294/pAmp streaked on the other LB plate.
11. Resterilize your loop as directed in step 5.
12. Repeat step 6 with *E. coli* MM294/pAmp streaked on the other LB/amp plate.
13. Sterilize and cool your inoculating loop.
14. Place all four streaked plates in a stack and tape them together. Seal the edges of your plates with masking tape.
15. Place the streaked plates upside down in the incubator. Alternatively, if you do not have an incubator, place the plates in a warm part of the room for a couple of days.

INVESTIGATION 20.1 *continued*

16. Disinfect your laboratory bench using the bleach solution.
17. Wash your hands thoroughly with soap and water.

Analysis

(a) After sufficient time has elapsed, remove your plates from the incubator and note any changes.

Never open the plates, as any bacterial colonies within are a potential source of contamination. If condensation has accumulated on one side of a plate, try looking through its bottom to observe the colonies you may have cultured. Once the experiment has been completed, flood plates with bleach to kill the bacterial colonies that have been cultured. Alternatively, place plates in an autoclave before they are disposed.

Evaluation

(b) Compare your results to your prediction. Explain any possible causes for variation.

(c) What evidence is there to indicate that protein was synthesized by the bacteria?

(d) Why was it important to streak out both types of bacteria on both types of plates?

(e) This experiment contains both positive and negative controls. Identify them. What information do the controls provide in this experiment?

(f) Why was it important to cool the inoculating loop before obtaining a bacterial colony from a stock plate?

(g) Why was it important to resterilize the inoculating loop between transfers of bacteria?

(h) Suggest possible sources of error in this procedure and indicate their effect on the results.

Synthesis

(i) *E. coli* strains containing the genetic sequence pAmp are resistant to ampicillin. Research how the ampicillin can be deactivated by β-lactamase, the protein coded for by the ampicillin-resistance gene.

(j) Predict what would happen if there was an error in the genetic sequence that codes for β-lactamase.

INVESTIGATION 20.2

Restriction Enzyme Digestion of Bacteriophage DNA

Report Checklist

● Purpose	● Design	● Analysis
○ Problem	○ Materials	● Evaluation
● Hypothesis	○ Procedure	○ Synthesis
● Prediction	● Evidence	

In this investigation, bacteriophage lambda DNA will be digested using the restriction endonucleases *Eco*RI, *Hin*dIII, and *Bam*HI. The fragments produced will be separated using gel electrophoresis. Fragment sizes will be calculated from an analysis of the agarose gel. Bacteriophage lambda DNA is obtained from a virus that infects bacterial cells and is 48 514 base pairs in length.

Before you begin, predict the number and size of the DNA fragments you will obtain, using the restriction enzyme site map shown in **Figure 1** on the next page.

Problem

How do the patterns of DNA fragments compare when a piece of DNA is digested using different restriction endonucleases?

Materials

safety goggles
gloves
70 % ethanol solution (or 10 % bleach)
4 1.5 mL Eppendorf tubes
waterproof pen for labelling
masking tape
polystyrene cup
freezer
crushed ice
20 μL of 0.5 μg/μL lambda DNA
5 μL 10× restriction buffer
1.0–20 μL micropipette with tips
2 μL each of *Bam*HI, *Eco*RI, and *Hin*dIII restriction endonucleases

INVESTIGATION 20.2 *continued*

microcentrifuge (optional)
37 °C water bath
thermometer
1 g agarose
paper boat
electronic balance
500 mL Erlenmeyer flask
250 mL graduated cylinder
microwave or hot plate
flask tongs or oven mitts
gel casting tray and gel electrophoresis box
1L 1× TBE buffer
5 µL loading dye
power supply (45 V)
plastic wrap
25–30 mL 0.025 % methylene blue, or enough to cover the gel in the staining tray
light box or overhead projector
acetate sheet

Wear safety goggles at all times.
Wear gloves when performing the experiment.
Wipe down all surfaces with 70 % ethanol, or 10 % bleach, before and after the laboratory exercise.
Do not use ethanol near a heat source.
Wash your hands thoroughly at the end of the laboratory.

Procedure

Day 1: Restriction Enzyme Digestion

1. Put on your safety goggles and gloves, and wipe down your bench with a 70 % ethanol solution (or 10 % bleach).

Ethanol is highly flammable. Make sure that any flame on your desk or near it is turned off before use.

2. Label four 1.5 mL Eppendorf tubes "*Bam*HI," "*Eco*RI," "*Hin*dIII," and "control." Place the tubes in a polystyrene cup containing crushed ice. **Table 1** outlines the amount of reagents to add to each tube. To keep track of each tube's contents, copy the table into your notebook and check off each reagent as you add it to the tube.

Table 1 Reagents to Add to Tubes

Tube	DNA (µL)	10× buffer (µL)	Water (µL)	*Bam*HI (µL)	*Eco*RI (µL)	*Hin*dIII (µL)
***Bam*HI**	4	1	4	1	–	–
***Eco*RI**	4	1	4	–	1	–
***Hin*dIII**	4	1	4	–	–	1
control	4	1	5	–	–	–

3. Read down each column, adding the same reagent to all appropriate tubes. Use a fresh tip on the micropipette for each reagent. Add the 4 µL of DNA to each tube first, followed by the 10× reaction buffer, and then the water. *Make sure you add the enzyme last.* Dispense all the contents close to the bottom of the Eppendorf tubes. Ensure that the pipette tip is touching the side of the tubes when dispensing the contents. *Keep everything on ice at all times.*
4. Close the Eppendorf tube tops. Place the tubes in the microcentrifuge, close it, and spin at maximum speed for approximately 3 s. If you do not have access to a microcentrifuge, then just tap the tubes on a soft pad or thick paper towel on the bench, pooling the contents to the bottom.

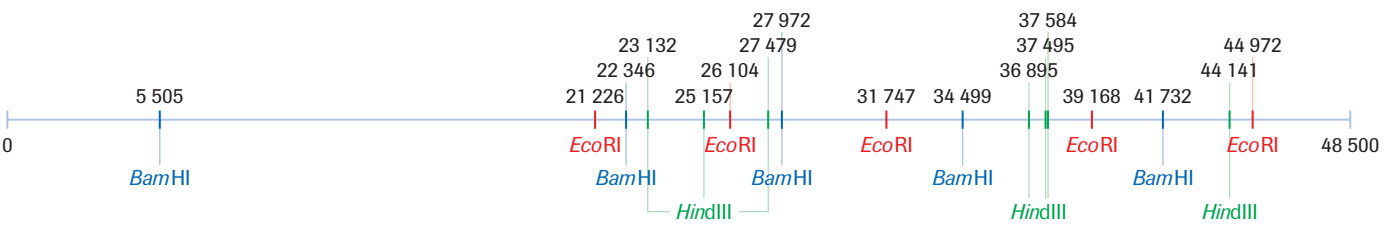

Figure 1
Restriction enzyme map of bacteriophage lambda DNA

INVESTIGATION 20.2 *continued*

When using the microcentrifuge:

- **Do not open the centrifuge until it stops completely.**
- **If the centrifuge tubes are smaller than the metal holder or holes, use the proper adaptor to accommodate them.**
- **Do not unplug the centrifuge by pulling on the cord. Pull the plug.**

5. Place the tubes in a 37 °C water bath for a minimum of 45 min. Use a thermometer to check the temperature of the water.
6. Once the digestion is complete, place the tubes in the polystyrene cup and put the cup in a freezer until your next class. Make sure you have labelled your cup with your name.

Day 2: Gel Electrophoresis

7. Measure 0.96 g of agarose powder in a paper boat on an electronic balance and transfer to a 500 mL Erlenmeyer flask.
8. Use a graduated cylinder to add 125 mL of 1× TBE buffer and swirl to mix.
9. Heat the flask on a hot plate or in a microwave until the solution is completely clear. Handle carefully, using tongs or oven mitts. Make sure you wear goggles and a lab coat.

If the agarose gets too hot it may bubble over. Be sure to observe your Erlenmeyer flask throughout the heating process. If the agarose solution starts to bubble up the neck of the flask, remove it immediately from the heat source using an oven mitt or tongs. Handle all hot glassware with caution.

10. Prepare the gel casting tray. Depending on your gel electrophoresis unit, you may have to tape the gel casting tray. Ensure that the plastic comb is inserted properly.
11. Once the flask with agarose solution is cool enough to handle with bare hands, pour the mixture into the gel casting tray. The comb teeth should be immersed in about 6 mm of agarose. The gel should cover only about one-third of the height of the comb teeth. Use a micropipette tip to remove bubbles from the gel as soon as it is poured.
12. Allow the agarose to set for a minimum of 20 min. The gel will become cloudy as it solidifies.
13. Once the gel has set (you may test this by gently touching the lower righthand corner with your finger), flood the gel with 1× TBE running buffer and then pull out the comb gently without ripping any of the wells.
14. Orient the tray containing the gel in the gel electrophoresis box so that the wells made by the comb are at the end with the positive electrode.
15. Add 1× TBE buffer to the gel electrophoresis box until the buffer is approximately 5 mm above the gel. Place the gel electrophoresis box to the side.
16. Add 1 μL of loading dye to each of the Eppendorf tubes. Microfuge for 3 s.
17. Micropipette the full contents of one Eppendorf tube into a well on the gel. Do the same for each tube. Be sure to record the order in which you dispense the tubes. Steady the micropipette over each well using both hands.
18. Close the gel box and connect it to the power supply. If you are using a gel box that you made, set the voltage to 45 V dc and turn it on. Electrophorese for 12 h. Alternatively, if you have a stronger power supply or a store-bought electrophoresis unit, electrophorese at 110 V for 2.5 h.

When using the power supply:

- **Be sure the grounding pin in the power supply is not broken.**
- **Pull the plug, not the cord, when unplugging the power source.**
- **Do not let the wire leads connected to the electric power supply or batteries touch each other.**

19. Unplug the power supply and carefully remove the gel. Wrap the gel in plastic wrap and place it in the refrigerator for a maximum of one day.

Day 3: Staining the Gel

20. Unwrap the gel and place it in the staining tray.
21. Flood the gel with 0.025 % methylene blue solution. Let the gel sit in the solution for at least 20 to 25 min. Pour off the water and replace it with fresh water. Repeat this process three more times. Keep an eye on the intensity of the DNA bands. If you destain for too long, you may lose the smaller fragments.

INVESTIGATION 20.2 *continued*

If you do not destain for long enough, the whole gel remains blue and the fragments cannot be differentiated.

22. Place the destained gel on a light box or on an overhead projector.
23. Obtain a blank acetate sheet or plastic wrap and place it over the gel. Trace the pattern of bands onto the wrap or sheet. Be sure to draw a line where the bottom of each well starts.

Evidence

(a) Carefully measure the distance in millimetres that each band migrated from the well origin. Copy **Table 2** into your notebook and use it to record the distances.

Analysis

(b) Using the *Hin*dIII digestion as a marker, plot the distance travelled (*x*-axis) versus the fragment base-pair size (*y*-axis) on semilogarithmic paper. Please note that the 23 130-base-pair fragment and the 27 491-base-pair fragment do not resolve, but instead travel as one band. Therefore, take an average of their size for graphing purposes.

(c) Using interpolation, determine the fragment size of the bands produced by digestion with *Bam*HI and *Eco*RI. Enter your calculated base-pair fragment sizes into your table.

(d) Compare the calculated base-pair fragments to the actual base-pair fragments. Use the restriction enzyme map of bacteriophage lambda (**Figure 1**) to determine the size of the actual band fragments for each enzyme. Calculate the percentage error.

Evaluation

(e) What was the purpose of each tube? of the control?

(f) Why do the smaller bands migrate faster than the larger bands?

(g) Some bands that are close in size migrate together. What measures may be taken to resolve bands close in size?

(h) What purpose does the 1× running buffer serve?

(i) Why must the gel be made using 1× TBE buffer?

(j) During electrophoresis, bubbles are produced at the anode and at the cathode. Explain why bubbles appear.

(k) Why must loading dye be added to the samples before they are loaded into the wells of the gel?

(l) Notice on your gel that the larger fragments are stained darker than the smaller fragments. Explain why this is the case.

(m) Suggest possible sources of error in this procedure. Indicate the effects of these sources of error on the results.

Table 2 Distance Travelled by Each Band From the Well Origin

*Hin*dIII		*Eco*RI			*Bam*HI		
Actual fragment size	***Distance travelled (mm)***	***Actual fragment size***	***Distance travelled (mm)***	***Calculated fragment size***	***Actual fragment size***	***Distance travelled (mm)***	***Calculated fragment size***
27 491							
23 130							
9 416							
6 557							
4 361							
2 322							
2 027							

Chapter 20 SUMMARY

Outcomes

Knowledge

- describe, in general, how genetic information is contained in the sequence of bases in DNA molecules in chromosomes; how the DNA molecules replicate themselves; and how the genetic information is transcribed into sequences of bases in RNA molecules and is finally translated into sequences of amino acids in proteins (20.1, 20.2)
- explain, in general, how restriction enzymes cut DNA molecules into smaller fragments and how ligases reassemble them (20.3)
- explain, in general, how cells may be transformed by inserting new DNA sequences into their genomes (20.3)
- explain how a random change (mutation) in the sequence of bases results in abnormalities or provides a source of genetic variability (20.4)
- explain how sequences of nucleic acids contained in the nucleus, mitochondria, and chloroplasts gives evidence for the relationships among organisms of different species by examining similarities and differences in base sequences (20.4)

STS

- explain that science and technology have both intended and unintended consequences for humans and the environment (20.3, 20.4)
- explain that scientific research and technological development help achieve a sustainable society, economy, and environment (20.3, 20.4)

Skills

- ask questions and plan investigations (20.4)
- conduct investigations and gather and record data and information (20.2, 20.3, 20.4)
- analyze data and apply mathematical and conceptual models to develop and assess possible solutions (20.2, 20.4)
- work as members of a team and apply the skills and conventions of science (all)

Key Terms

20.1

complementary base pairing
antiparallel
DNA replication
semiconservative replication
template
DNA helicase
DNA polymerase III
leading strand
lagging strand
DNA polymerase I
DNA ligase

20.2

gene expression
ribonucleic acid (RNA)
transcription
messenger RNA (mRNA)
translation
RNA polymerase
promoter
template strand
termination sequence
codon
start codon
stop codon
ribosome
transfer RNA (tRNA)
anticodon

20.3

recombinant DNA
genetic transformation
restriction endonuclease
recognition site
palindromic
sticky ends
blunt ends
methylase
polymerase chain reaction (PCR)
vector
transgenic
plasmid
multiple-cloning site

20.4

point mutation
gene mutation
silent mutation
missense mutation
nonsense mutation
deletion
insertion
frameshift mutation
translocation
inversion
spontaneous mutation
mutagenic agent
induced mutation
phylogeny
SINEs
LINEs

MAKE a summary

1. Starting with the title "The Human Genome," produce a flowchart that illustrates the flow of information from gene to protein. Include as many key concepts as possible.
2. Revisit your answers to the Starting Points questions at the beginning of the chapter. Would you answer the questions differently now? Why?

Go To

www.science.nelson.com GO

The following components are available on the Nelson Web site. Follow the links for *Nelson Biology Alberta 20–30*.

- an interactive Self Quiz for Chapter 20
- additional Diploma Exam-style Review Questions
- Illustrated Glossary
- additional IB-related material

There is more information on the Web site wherever you see the Go icon in the chapter.

EXTENSION

Cracking the Code of Life

In this *NOVA* video, follow corporate and academic scientists as they race to capture one of the biggest prizes in scientific history: the complete, letter-by-letter sequence of genetic information that defines human life—the human genome.

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EXTENSION

Artificial Life

Scientists can now synthesize strands of DNA with any nucleotide sequence they want. Does this mean that they can create artificial life from these blueprints? Some scientists believe the answer is yes, and that it isn't that far away! Find out why by watching this *NOVA* video.

www.science.nelson.com GO

EXTENSION

CBC radioONE QUIRKS & QUARKS

DNA Motors

Dr. Vanessa Auld, Quirks and Quarks genetics columnist explains the details behind the discovery by a group of American and Czech researchers of proteins that act like small motors inside the nucleus of the cell. This discovery is changing our understanding of how DNA is used to manufacture the proteins and chemicals the cell uses to sustain life.

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EXTENSION

Golden Rice or Frankenfood?

Vitamin A deficiency is a leading cause of preventable blindness. Scientists have developed a genetically-modified rice that contains β-carotene, the precursor to vitamin A. Some see this new rice as an important contribution to world health, but others warn that genetically modified foods could have hidden dangers. What do you think?

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UNIT 30 C PERFORMANCE TASK

Investigating Human Traits

In this Performance Task, you will use the skills you gained in this Unit to design and carry out a correlational study on human traits to determine if they are autosomal or sex-linked. Go to the Unit 30 C Performance Task link on the Nelson web site to complete the task.

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Chapter 20 REVIEW

Many of these questions are in the style of the Diploma Exam. You will find guidance for writing Diploma Exams in Appendix A5. Science Directing Words used in Diploma Exams are in bold type. Exam study tips and test-taking suggestions are on the Nelson Web site.

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DO NOT WRITE IN THIS TEXTBOOK.

Part 1

Use the following information to answer questions 1 to 3.

The cause of cystic fibrosis has been identified as a variety of mutations to the *CFTR* gene on chromosome 7. The most common of these involves the loss of three nucleotides, which in turn results in the loss of a phenylalanine at amino acid position 508.

1. Identify the DNA sequence that would result in phenylalanine being placed in a polypeptide chain.
   A. UUG
   B. AAC
   C. UUU
   D. TTT

2. Identify the term that best describes the mutation that causes the loss of phenylalanine.
   A. silent mutation
   B. insertion mutation
   C. deletion mutation
   D. missense mutation

3. NR Gene therapy trials to correct this defect in the *CFTR* gene have been conducted by doctors in several centres. The following is a list of some genetic technologies that might be used in this work:
   1. restriction endonucelases
   2. mtDNA
   3. polymerase chain reaction
   4. DNA ligase
   5. viruses
   6. bacterial plasmids
   7. gene sequencing

   Identify the technologies that would most likely be used to isolate the gene for a therapy trial. (Record all four digits of your answer in the order in which the technologies would be used.)

4. Identify the enzyme that is correctly matched with its function.
   A. DNA polymerase I: synthesis of the continuous matching strand
   B. DNA helicase: synthesis of messenger RNA
   C. DNA polymerase III: cuts out the primer and replaces it with DNA nucleotides
   D. DNA ligase: links adjacent nucleotides together by covalent bond

5. Select the response that correctly identifies the complementary DNA strand for this strand:
5′-TACTTTGGCCCAGAG-3′
   A. 3′-AUGAAACCGGGUCUC-5′
   B. 3′-UACUUUGGCCCAGA-5′
   C. 3′-ATGAAACCGGGTCTC-5′
   D. 5′-ATGAAACCGGGTCTC-3′

Use the following information to answer questions 6 and 7.

1. Amino acids are brought to the ribosome and linked together in the correct order.
2. A copy of the gene is taken to the ribosome.
3. RNA polymerase attaches to the promoter site.
4. The two subunits of the ribosome attach to the RNA strand.
5. DNA polymerase III makes a matching strand using complementary base pairs.
6. Release factor binds to the A site and the ribosome releases the amino acid chain.
7. The two original strands serve as templates for the synthesis of new matching stands.
8. The lagging strand is synthesized in short fragments.
9. The two strands are unwound and the hydrogen bonds are broken.

6. NR Identify the steps described above that correspond to the process of replication. (Record all four digits of your answer in the order the steps would occur in the cell.)

7. NR Match these terms to the selection above that best describes them. (Record all four digits of your answer.)

_____	_____	_____	_____
initiation of transcription	termination of translation	elongation of amino acid chain	initiation of translation

Part 2

8. Use a diagram to **illustrate** how the two DNA strands in a double helix run antiparallel. Make sure you label your diagram.

9. **How** does the fact that DNA replicates semiconservatively decrease the possibility of errors made during DNA replication? **Describe** another mechanism that minimizes DNA replication error.

10. Numerous enzymes are involved in DNA replication. **Outline** the role that the following enzymes play: DNA ligase, DNA gyrase, DNA helicase, DNA polymerase I, and DNA polymerase III.

11. What is the complementary strand of AATTGCATA?

12. DNA polymerase III can only extend an existing DNA strand in the 5′ to 3′ direction. **Describe** the mechanisms in place that compensate for DNA polymerase III's inability to intitiate a strand and for its stringent directionality.

13. One strand of a DNA molecule contains the nucleotide proportions 15 % adenine (A), 30 % thymine (T), 20 % guanine (G), and 35 % cytosine (C). **Predict** the proportions of the four base pairs in the double-stranded form of this DNA.

14. **Describe** the function of mRNA and tRNA in protein synthesis.

15. **Distinguish** between transcription and translation. Use a table to organize your answer.

16. The following is a sequence of DNA for a hypothetical peptide:
5′- AAGTACAGCAT - 3′
3′- TTCATGTCGTA - 5′
Translate this sequence into protein using the genetic code.

17. Every codon consists of a triplet of base pairs. **Explain** why amino acids cannot be coded with just two base pairs.

18. **Describe** how the structure of mRNA is similar to DNA. **How** does mRNA differ from DNA?

19. DE Cutting a piece of DNA with a restriction enzyme can give DNA fragments with sticky ends or with blunt ends, depending on the restriction enzyme that is used. Write a unified response addressing the following aspects of cutting DNA with a restriction enzyme:
    - **Distinguish** between sticky ends and blunt ends.
    - **Describe** how a DNA fragment with a sticky ends could be produced.
    - **Describe** how a DNA fragment with blunt ends could be produced.
    - **Illustrate** your descriptions with diagrams.

20. The DNA fragment CGTCATCGATCATGCAGCTC contains a restriction enzyme recognition site. **Identify** the site.

21. **Explain** how the presence of an antibiotic-resistance marker gene in a plasmid can be used to determine whether a transformation protocol has been successful.

22. Recently, the Human Genome Project (HGP) was completed. The HGP has provided us with a complete sequence of the human genome. Despite this great advancement, we are far away from realizing the numerous medical treatments that will eventually be made available because of, or as a result of, the project. Scientists are now working on the Human Proteome Project, which involves linking genes to both functional and dysfunctional proteins. **Explain** why there would be limited progress in medical research if scientists were restricted to working only with DNA sequences and not with proteins.

23. DE *Pseudomonas syringae* is a bacterium found in raindrops and most ice crystals. These bacteria act as nuclei for ice crystal formation, catalyzing ice formation at temperatures approaching 0 °C. It does so by producing an ice-nucleation protein in the outer membrane of its cells. Researchers have been able to cleave the gene for this protein from its genome, thereby preventing the bacteria from forming ice crystals. When the genetically engineered "ice-minus" bacteria are sprayed on tomato plants, frost damage is reduced. The presence of the ice-minus bacteria can extend growing seasons, thus increasing crop yields, especially in cold climates. However, environmental groups have raised serious concerns about releasing genetically engineered bacteria into the environment. Write a unified response addressing the following aspects of the use of ice-minus bacteria:
    - **Predict** whether the new microbes could gain a selective advantage over the naturally occurring species?
    - **Describe** what might happen if the genetically engineered microbes mutate?
    - Do you think that genetically engineered microbes should be introduced into the environment? **Justify** your opinion.

Use the following information to answer questions 24 to 26.

Huntington disease is an inherited disorder that manifests itself in abnormal body movements and memory loss that degenerates into dementia and cognitive decline. This disorder is caused by a codon repeat in the Huntington protein gene on chromosome 4. In the normal form of this gene there are fewer than 40 repeats of the codon CAG. More repeats result in the eventual onset of the disease and severity seems to increase with the number of repeats.

24. **Identify** the amino acid specified by the CAG codon.

DE

25. **Explain** what increased inclusions of the CAG codon in the Huntington gene might do to the protein structure of the Huntington protein.

DE

26. **Describe** the steps a lab would take to diagnose the number of CAG repeats on the Huntington gene of an individual. **Identify** the specific technologies and **describe** how they would be used in this analysis.

DE

Use the following information to answer questions 27 and 28.

The first recombinant DNA organisms were bacteria that were altered for commercial purposes to produce a protein product when grown in culture. These recombinant organisms caused little public concern, as they were perceived to be contained within a laboratory or factory. However, subsequent genetic engineering projects have included the release of engineered organisms into the environment. Agricultural transgenic products being grown today include golden rice, insect-resistant maize and cotton, and herbicide-resistant canola and corn, to name a few.

27. **Identify** and **describe** the technologies used to create these recombinant organisms.

28. **Explain** the concerns of those who oppose the use of these organisms, and the benefits touted by their proponents.

Use the following information to answer questions 29 to 32.

A company, Gene Tree, offers kits that can be used to test whether an individual has DNA sequences found most often in the Aboriginal peoples. The tests are compared to known genetic markers in mitochondrial DNA (mtDNA), Y chromosome DNA, and nuclear DNA that are unique to the Aboriginal peoples of North America. Testing to establish the ethnic background of individuals may raise concerns about the use of this information. Historically, there have been political, legal, and moral issues around attempts to identify an individual's ethnicity as distinct from those of others.

29. **Identify** the technique listed above that would best determine paternal inheritance of Aboriginal ancestry. **Explain** your selection.

30. **Sketch** a diagram of meiosis that shows the formation of a human egg. Label the important features and clearly label the ploidy of the key stages. Describe how mtDNA is inherited during the formation of the human zygote, and identify which parent would be contributing the genetic markers for Aboriginal ancestry if they were identified in mtDNA.

31. **Identify** and **describe** two DNA technologies that would be used to carry out these tests.

32. **Identify** two advantages and two disadvantages to both society and individuals that might arise from using DNA technology to trace ethnic patterns of inheritance.

Many of these questions are in the style of the Diploma Exam. You will find guidance for writing Diploma Exams in Appendix A5. Science Directing Words used in Diploma Exams are in bold type. Exam study tips and test-taking suggestions are on the Nelson Web site.

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DO NOT WRITE IN THIS TEXTBOOK.

Part 1

1. NR Indicate the correct order, beginning with prophase, of the following events of cell division.

1. Nuclear membrane begins to dissolve.
2. Chromatids move to opposite poles.
3. Chromosomes align along the equatorial plate.
4. Chromosomes reach opposite poles and begin to lengthen.

2. A fertilized mosquito egg has six chromosomes. During mitosis, the egg cell undergoes multiple divisions. Which row shows the correct number of chromosomes found in telophase and interphase?

	Number of chromosomes	
Row	**Telophase**	**Interphase**
A.	3	3
B.	3	6
C.	6	3
D.	6	6

Use the following information to answer questions 3 and 4.

Figure 1 shows four phases of cell division in a plant cell.

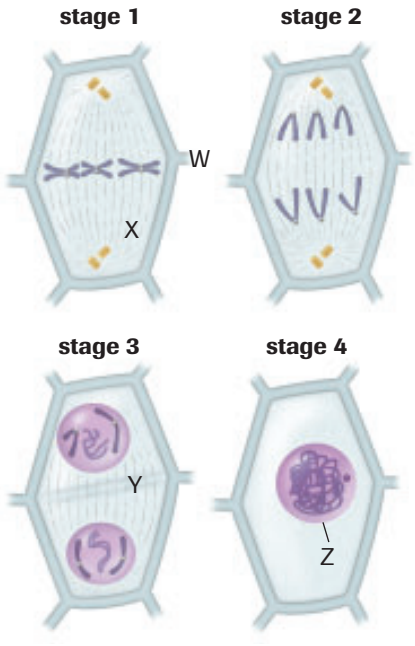

Figure 1

3. NR Identify the phases of cell division in **Figure 1**.

_____ prophase _____ metaphase _____ anaphase _____ telophase

4. The correct labels for the structure identified by letters in **Figure 1** are

A. W = centriole, X = centromere, Y = cytoplasm, Z = nucleolus
B. W = centromere, X = spindle fibre, Y = division plate, Z = nuclear membrane
C. W = chromatid, X = centromere, Y = nuclear membrane, Z = nucleolus
D. W = chromosome, X = spindle fibre, Y = chromatin, Z = nuclear membrane

Use the following information to answer questions 5 to 7.

A corn plant with white seeds, a large cob, and small leaves is crossed with a corn plant with yellow seeds, a large cob, and large leaves. All of the F_1 offspring have yellow seeds, large cobs, and large leaves.

5. Identify the row that correctly gives the dominant traits, according to this data.

Row	**Seed colour**	**Cob type**	**Leaf size**
A.	white	large	small
B.	white	small	large
C.	yellow	small	small
D.	yellow	large	large

6. Identify the row that correctly gives the expected traits of the offspring, if a plant from the F_1 generation were cloned.

Row	**Seed colour**	**Cob type**	**Leaf size**
A.	white	large	small
B.	white	small	large
C.	yellow	small	small
D.	yellow	large	large

7. If a plant from the F_1 generation were crossed with a corn plant with white seeds, identify the colour seeds you would expect to see in the F_2 generation.

A. 100 % of individuals would have white seeds
B. 75 % of individuals would have yellow seeds and 25 % white seeds
C. 50 % of individuals would have white seeds and 50 % yellow seeds
D. 100 % of individuals would have yellow seeds

8. Correctly match the cell number in **Figure 2** with the condition. (Record all four digits of your answer.)

NR

_____	_____	_____	_____
Turner's syndrome	trisonomic female	will not survive	trisonomic male

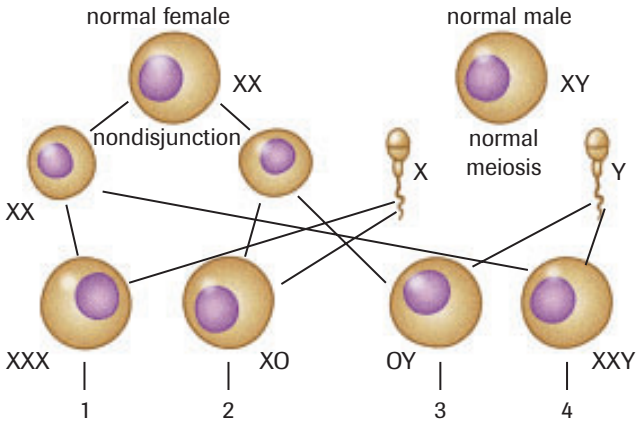

Figure 2

Use the following information to answer questions 9 and 10.

Thalassemia is a serious human genetic disorder that causes severe anemia. People with thalassemia die before sexual maturity. There are over 90 different mutations that can lead to thalassemia. One of the mutations changes the codon TAC to TAA.

9. Identify the row that best describes the type of mutation and its consequence to the structure of the protein.

Row	Mutation	Consequence
A.	insertion	causes a shift to the reading frame and results in an entirely different amino acid sequence
B.	deletion	causes a shift to the reading frame and results in an entirely different amino acid sequence
C.	substitution	causes a different amino acid at one location
D.	inversion	causes different amino acids for the sequence inverted as reading frame is reversed

10. Select the statement that best describes the codons given in the description of thalassemia.

A. These are mRNA codons as they contain the base uracil.
B. These are mRNA codons as they contain the base thyamine.
C. These are DNA codons as they contain the base uracil.
D. These are DNA codons as they contain the base thymine.

Use the following information to answer questions 11 and 12.

1. Initiation commences when the RNA polymerase binds to the promoter region of the gene to be transcribed.
2. The ribosome continues to move along the mRNA, reading the code in triplets known as codons.
3. When the ribosome moves over, the tRNA containing the growing peptide is shifted over to the P site. A third amino acid, specified by the third codon, is brought into the A site by the next tRNA. A peptide bond is formed between the second and third amino acid.
4. A complementary RNA strand is synthesized in the direction of 5′ to 3′, using one strand of DNA as a template. This step is known as elongation. The complement of adenine in RNA is uracil.
5. New amino acids are added to the chain in the process of elongation, which continues until a stop codon is read in the A site. The stop codons are UAG, UGA, and UAA. At this point, the ribosome stalls.
6. Once the termination sequence is reached by the RNA polymerase, the process ceases. The mRNA is separated from the DNA and the RNA polymerase falls off the DNA molecule.
7. When the start codon is in the P site, a tRNA delivers the amino acid methionine. The tRNA recognizes the codon because of the complementary anticodon.

11. Identify the statements that describe the process of transcription. (Record all three digits of your answer in the order in which they would occur in the cell.)

NR

12. Identify the statements that describe the process of translation. (Record all four digits of your answer in the order in which they would occur in the cell.)

NR

Use the following information to answer questions 13 to 15.

Genetic inheritance of risk for certain types of breast cancer has long been inferred from its incidence in family clusters. Mutations in either the *BRCA*1 or *BRCA*2 genes accounts for 2 % to 3 % of breast cancers and 9 % of ovarian cancers. People who are identified as having a mutation in either of these genes have a 60 % to 85 % lifetime risk of getting breast cancer and a 15 % to 40 % lifetime risk of getting ovarian cancer. The gene *BRCA*1 is located on chromosome 17 and codes for approximately 1800 amino acids, while the gene *BRCA*2 is located on chromosome 13 and codes for approximately 3400 amino acids.

13. Select the statement that is supported by these data.

A. Mutations in the *BRCA*1 and *BRCA*2 genes are inherited in an autosomal recessive pattern.

B. Mutations in the *BRCA*1 and *BRCA*2 genes always cause breast cancer and sometimes cause ovarian cancer.

C. Mutations in the *BRCA*1 and *BRCA*2 genes cause cancer when influenced by environmental factors.

D. Mutations in the *BRCA*1 and *BRCA*2 genes always cause ovarian cancer and sometimes cause breast cancer.

14. NR Determine the minimum number of base pairs a *BRCA*2 gene would contain to code for a complete protein. (Record all four digits of your answer.)

15. Identify which of the following statements about *BRCA*1 and *BRCA*2 gene mutations is incorrect:

A. A woman's risk for genetically linked breast cancer is only elevated if the maternal branch of her family had a history of breast cancer.

B. Mutations in the *BRCA* genes also increase the risk of ovarian cancer.

C. Women without mutations in the *BRCA* genes may still be at high risk of getting breast cancer.

D. A woman's lifetime risk of genetically linked breast cancer is elevated if there is a family history of breast cancer in either branch of her family.

Part 2

16. Genetic testing to identify mutations of the *BRCA*1 and *BRCA*2 genes can be accomplished by gene cloning. **Explain** why a patient might or might not want to have such genetic tests done.

Use the following information to answer questions 17 to 19.

A student observed fertilized eggs of two different species, whitefish and frog, undergoing mitosis. The number of cells in each stage of the cell cycle, at the time the egg masses were prepared and mounted on a slide, were counted. These numbers are presented in **Table 1**.

Table 1 Number of Cells in Specific Stages of the Cell Cycle

Cell cycle stage	Whitefish	Frog
interphase	81	88
prophase	10	6
metaphase	5	5
anaphase	1	0
telophase	3	1

17. DE **Determine** the total time for which cells were in each phase, for both whitefish and frog cells.

18. DE **Identify** the phase of the cell cycle that took the longest time to complete, for both whitefish and frog cells.

19. DE **Sketch** the cell cycle for the fertilized whitefish and frog eggs.

Use the following information to answer questions 20 to 22.

Cancer cells can divide at rates that far exceed those of normal cells. Some drugs used to treat cancer block the action of enzymes that are essential for chromosomal duplication.

20. DE **Why** would these drugs be useful in treating cancer?

21. DE **Predict** the phase of the cell cycle that would likely be affected by these drugs.

22. DE **Predict** the phase of mitosis that might be affected.

23. Approximately 25 plant species make up about 90 % of the human diet. Some scientists have speculated that global warming will reduce plant diversity, making us even more dependent on these species. Our ability to maintain our food supply under these conditions would require advances in genetic engineering, selective breeding, and cloning of plants. **Describe** ways in which these technologies might be used to increase crop production.

Use the following information to answer questions 24 to 27.

Figure 2 is a flowchart showing the stages of meiosis for spermatogenesis.

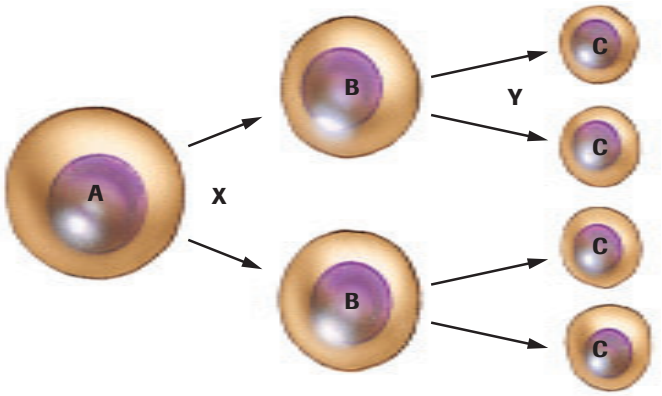

Figure 2

24. **Identify** the stage of meiosis indicated by the labels X and Y on **Figure 2**.

25. **Describe** the events at each stage of meiosis shown on the flowchart in **Figure 2**.

26. **Identify** which cells in **Figure 2** would have haploid chromosomes?

27. Cell A in **Figure 2** contains 44 chromosomes. **Infer** the number of chromosomes that cell C contains.

Use the following information to answer questions 28 to 30.

A normal human sperm cell fertilizes an egg cell containing 24 chromosomes. A lab technician examining a karotype of fetal cells notices trisomy of chromosome pair 21.

28. **Sketch** the karotype.

29. **Predict** how many chromosomes will be found in a muscle cell of the fetus.

30. From the information provided, is it possible to predict the sex of the embryo? **Explain** your answer.

31. Gene therapy is a technique in which defective genes are located and substituted by normally functioning genes. In the future, gene banks may likely be a common source of genes for treating genetic disorders. **List** two potential disadvantages to society of the use of gene banks.

Use the following information to answer questions 32 to 34.

Figure 3 shows the formation of sex cells in a mammal.

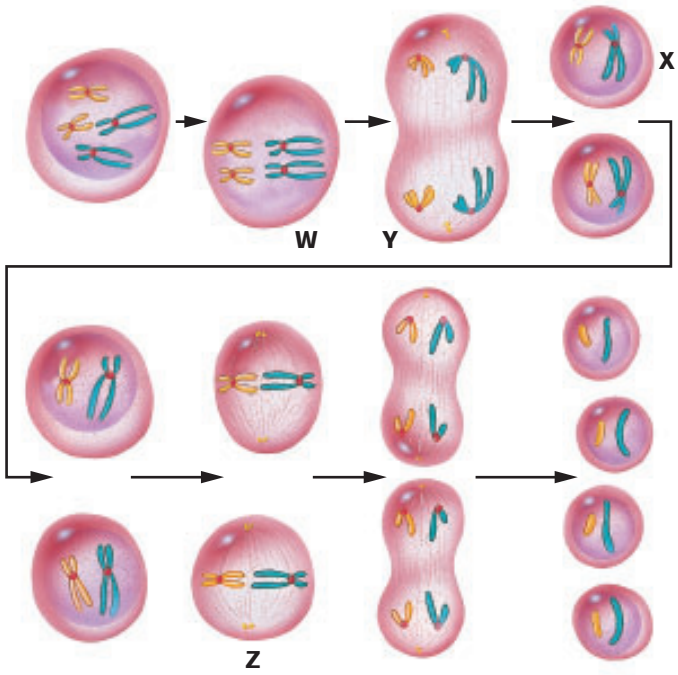

Figure 3

32. Use the diagrams and labels in **Figure 3** to help **explain** the process of crossing over.

33. **Identify** correctly the letter label in **Figure 3** that marks the first haploid cells formed by meiosis. Explain your answer.

34. **Identify** the correct letter label that marks metaphase II. What is happening during this phase?

Use the following information to answer questions 35 and 36.

In guinea pigs, black hair is dominant to white hair, and short hair is dominant to long hair. A guinea pig that is homozygous for both white hair and for short hair is mated with a guinea pig that is homozygous for both black hair and for long hair.

35. **Predict** the phenotype(s) of the F_1 generation.

36. Two members of the F_1 generation are mated. **Determine** the predicted phenotype ratio for the F_2 generation.

37. In chickens, the allele for rose comb (*R*) is dominant over the allele for single comb (*r*), and the allele for feathered legs (*F*) is dominant to the allele for clean legs (*f*). A breeder mates four birds with feather legs and rose combs. The phenotypes of the offspring of these crosses are shown in **Table 2**. **Determine** the genotypes of the parents.

Table 2

Parents	Phenotype of F_1 offspring
rooster A → hen C	all have rose combs; some have feathered legs and some have clean legs
rooster A → hen D	all rose combs and feathered legs
rooster B → hen C	most have rose combs, some have single combs; all have feathered legs
rooster B → hen D	rose and single combs; all have feathered legs

38. In mice, coat colour is determined by more than one gene. For one gene, the allele *C* determines a coloured coat, and the allele *c* determines an albino phenotype. For a second gene, the *B* allele causes activation of a pigment that produces black coat colour. The recessive allele, *b*, causes incomplete activation of the pigment, producing brown coat colour. These two genes are located on separate chromosomes and segregate independently. **Determine** the predicted genotypic and phenotypic ratios of the F_1 generation from the cross *CcBb* × *CcBb*.

39. In your notebook, construct a table to **compare** replication, transcription, and translation. (A comparison includes similarities and differences.) Your table should include the following headings: Process name, Location in cell, Time during cell cycle, Product, Brief summary of process.

40. In actively dividing cells, DNA replication occurs during interphase. **Sketch** the process of replication, using the following segment of DNA as an example:

5′-AAAAATTTAATATATTACAATGGCCCCGCGAT
AGTTCGTAGT-3′

3′-TTTTTAAATTATATAATGTTACCGGGGCGCTAT
CAAGCATCA-5′

Label and annotate your diagram to describe the process. Clearly indicate the start codon on your diagram.

Use the following information to answer questions 41 to 44.

Tay Sach disease results from a mutation in the gene for the enzyme hexoseaminidase. This mutation is an autosomal recessive disorder. The absence of a correct gene for this enzyme results in an inability to break down fatty material called ganglioside, which causes eventual death as the ganglioside builds up in the brain. There is no effective treatment for this disease.

5′-AUGCAGGUGACCUCAGUG-3′
mRNA sequence for normal protein

5′-AUGCAGGUGACAUACCUCAGUG-3′
mRNA sequence for mutated protein

41. DE Give the amino acid sequence that would result from translation of the mRNA at the ribosome.

42. DE Write the sequence for the normal and mutated protein into your notebook. **Determine** the DNA sequence from which each sequence is transcribed.

43. DE Tay Sach disease is the result of a gene mutation. **Identify** the mutation by circling the changed sequence. Name the type of mutation that has occurred and **explain** the changes that would occur in the protein.

44. DE **Outline** the procedure that you would follow to attempt a gene therapy treatment for Tay Sach disease. Start from the assumption you already know the sequence of the normal gene.

45. **Describe** an advantage and disadvantage to treating individuals with Tay Sachs by applying gene therapy to somatic cells. **Describe** an advantage and disadvantage to treating individuals with Tay Sachs by applying gene therapy to sex cells.

46. The gene for growth hormone has been isolated from human chromosomes and cloned in bacteria. The bacteria produce human growth hormone, which can be harvested in large quantities. The hormone is invaluable to people with dwarfism. Before its development, people with dwarfism relied on costly pituitary extracts. Although the prospect of curing dwarfism has been met with approval, some concerns have been raised about the potentially vast supply of growth hormone. Should individuals who do not have dwarfism but who want to grow a few more centimetres have access to the growth hormone biotechnology? **Justify** your opinion.

47. Review the focusing questions on page 552. Using the knowledge you have gained from this unit, briefly **outline** a response to each of these questions.