4.3 Powering the Cell: Cellular Respiration

Lesson Objectives

- Name the three stages of cellular respiration.
- Give an overview of glycolysis.
- Explain why glycolysis probably evolved before the other stages of aerobic respiration.
- Describe the mitochondrion and its role in aerobic respiration.
- List the steps of the Krebs cycle, and identify its products.
- Explain how electron transport results in many molecules of ATP.
- State the possible number of ATP molecules that can result from aerobic respiration.

Vocabulary

aerobic respiration type of cellular respiration that requires oxygen

anaerobic respiration type of cellular respiration that does not require oxygen

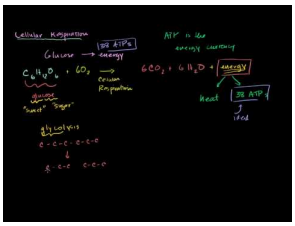
glycolysis first stage of cellular respiration in which glucose is split, in the absence of oxygen, to form two molecules of pyruvate (pyruvic acid) and two (net) molecules of ATP

Krebs cycle second stage of aerobic respiration in which two pyruvate (pyruvic acid) molecules from the first stage react to form ATP, NADH, and $FADH_2$

Introduction

You have just read how photosynthesis stores energy in glucose. How do living things make use of this stored energy? The answer is cellular respiration. This process releases the energy in glucose to make ATP, the molecule that powers all the work of cells.

An introduction to cellular respiration can be viewed at http://www.youtube.com/user/khanacademy#p/c/7A9646BC5110CF64/19/2f7YwCtHcgk (14:19).



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Stages of Cellular Respiration

Cellular respiration involves many chemical reactions. As you saw earlier, the reactions can be summed up in this equation:

$C_6H_{12}O_6 + 6O_2 \rightarrow 6CO_2 + 6H_2O$ + Chemical Energy (in ATP)

The reactions of cellular respiration can be grouped into three stages: glycolysis, the Krebs cycle (also called the citric acid cycle), and electron transport. **Figure** 4.10 gives an overview of these three stages, which are also described below.

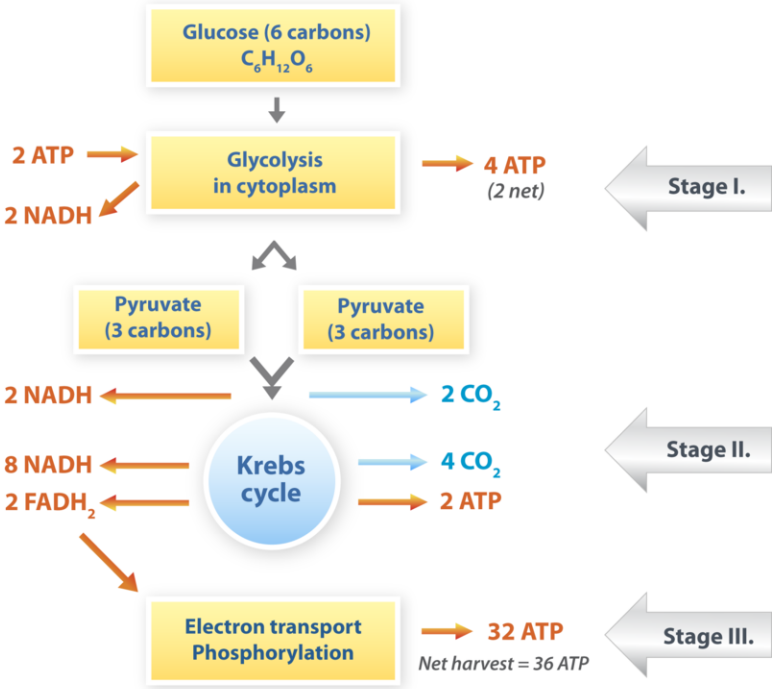


FIGURE 4.10

Cellular respiration takes place in the stages shown here. The process begins with a molecule of glucose, which has six carbon atoms. What happens to each of these atoms of carbon?

Cellular Respiration Stage I: Glycolysis

The first stage of cellular respiration is **glycolysis**. It takes place in the cytosol of the cytoplasm.

Splitting Glucose

The word *glycolysis* means "glucose splitting," which is exactly what happens in this stage. Enzymes split a molecule of glucose into two molecules of pyruvate (also known as pyruvic acid). This occurs in several steps, as shown in **Figure** 4.11. You can watch an animation of the steps of glycolysis at the following link: http://www.youtube.com/watch?v=6JGXayUyNVw.

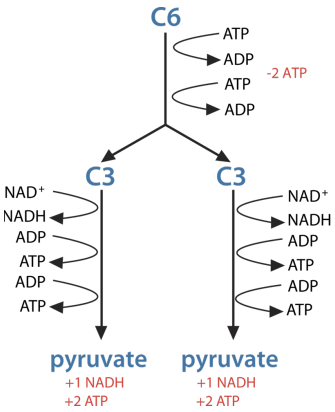


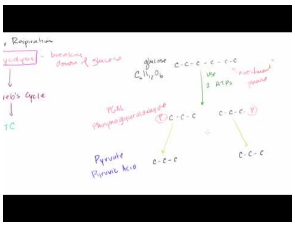
FIGURE 4.11

In glycolysis, glucose (C6) is split into two 3-carbon (C3) pyruvate molecules. This releases energy, which is transferred to ATP. How many ATP molecules are made during this stage of cellular respiration?

Results of Glycolysis

Energy is needed at the start of glycolysis to split the glucose molecule into two pyruvate molecules. These two molecules go on to stage II of cellular respiration. The energy to split glucose is provided by two molecules of ATP. As glycolysis proceeds, energy is released, and the energy is used to make four molecules of ATP. As a result, there is a net gain of two ATP molecules during glycolysis. During this stage, high-energy electrons are also transferred to molecules of NAD^+ to produce two molecules of NADH, another energy-carrying molecule. NADH is used in stage III of cellular respiration to make more ATP.

A summary of glycolysis can be viewed at http://www.youtube.com/user/khanacademy#p/c/7A9646BC5110CF64/22/FE2jfTXAJHg.



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Anaerobic and Aerobic Respiration

Scientists think that glycolysis evolved before the other stages of cellular respiration. This is because the other stages need oxygen, whereas glycolysis does not, and there was no oxygen in Earth's atmosphere when life first evolved about 3.5 to 4 billion years ago. Cellular respiration that proceeds without oxygen is called **anaerobic respiration**. Then, about 2 or 3 billion years ago, oxygen was gradually added to the atmosphere by early photosynthetic bacteria. After that, living things could use oxygen to break down glucose and make ATP. Today, most organisms make ATP with oxygen. They follow glycolysis with the Krebs cycle and electron transport to make more ATP than by glycolysis alone. Cellular respiration that proceeds in the presence of oxygen is called **aerobic respiration**.

Structure of the Mitochondrion: Key to Aerobic Respiration

Before you read about the last two stages of aerobic respiration, you need to know more about the mitochondrion, where these two stages take place. A diagram of a mitochondrion is shown in **Figure** 4.12.

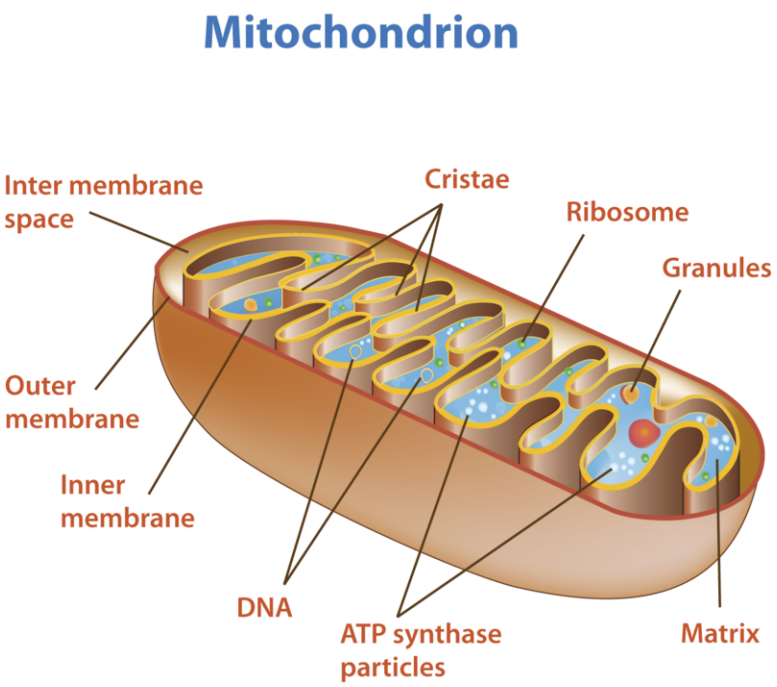


FIGURE 4.12

The structure of a mitochondrion is defined by an inner and outer membrane. This structure plays an important role in aerobic respiration.

As you can see from **Figure** 4.12, a mitochondrion has an inner and outer membrane. The space between the inner and outer membrane is called the intermembrane space. The space enclosed by the inner membrane is called the matrix. The second stage of cellular respiration, the Krebs cycle, takes place in the matrix. The third stage, electron transport, takes place on the inner membrane.

Cellular Respiration Stage II: The Krebs Cycle

Recall that glycolysis produces two molecules of pyruvate (pyruvic acid). These molecules enter the matrix of a mitochondrion, where they start the **Krebs cycle**. The reactions that occur next are shown in **Figure** 4.13. You can watch an animated version at this link: http://www.youtube.com/watch?v=p-k0biO1DT8&feature=related.

Before the Krebs cycle begins, pyruvic acid, which has three carbon atoms, is split apart and combined with an enzyme known as CoA, which stands for coenzyme A. The product of this reaction is a two-carbon molecule called acetyl-CoA. The third carbon from pyruvic acid combines with oxygen to form carbon dioxide, which is released as a waste product. High-energy electrons are also released and captured in NADH.

Steps of the Krebs Cycle

The Krebs cycle itself actually begins when acetyl-CoA combines with a four-carbon molecule called OAA (oxaloacetate) (see **Figure** 4.13). This produces citric acid, which has six carbon atoms. This is why the Krebs cycle is

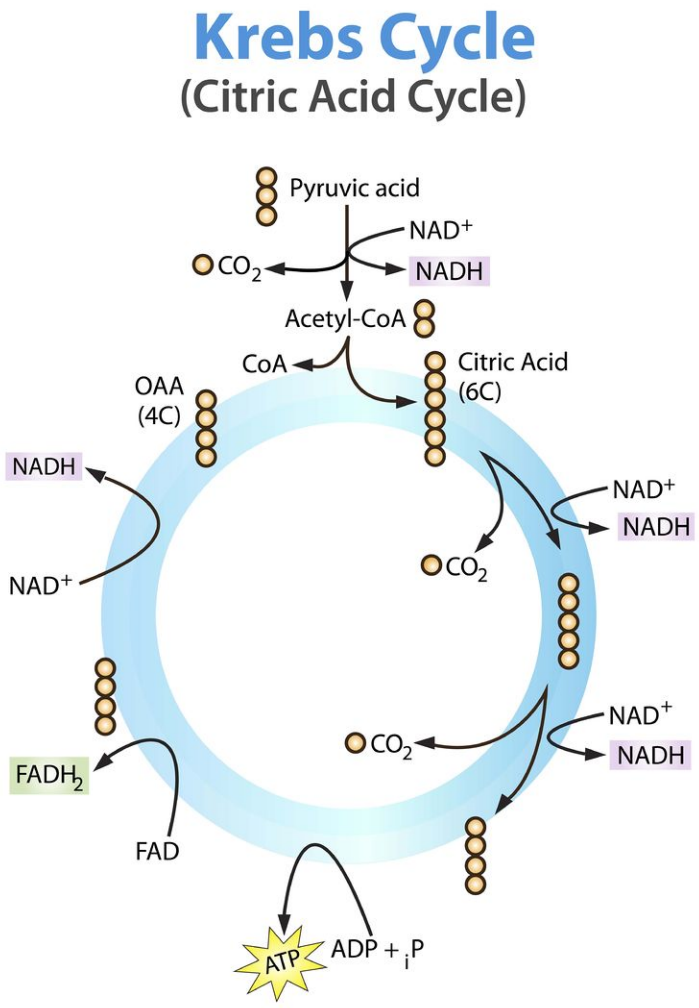


FIGURE 4.13

The Krebs cycle starts with pyruvic acid from glycolysis. Each small circle in the diagram represents one carbon atom. For example, citric acid is a six carbon molecule, and OAA (oxaloacetate) is a four carbon molecule. Follow what happens to the carbon atoms as the cycle proceeds. In one turn through the cycle, how many molecules are produced of ATP? How many molecules of NADH and FADH2 are produced?

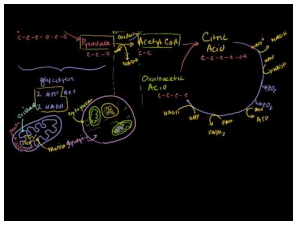
also called the citric acid cycle. After citric acid forms, it goes through a series of reactions that release energy. The energy is captured in molecules of NADH, ATP, and $FADH_2$, another energy-carrying compound. Carbon dioxide is also released as a waste product of these reactions. The final step of the Krebs cycle regenerates OAA, the molecule that began the Krebs cycle. This molecule is needed for the next turn through the cycle. Two turns are needed because glycolysis produces two pyruvic acid molecules when it splits glucose. Watch the OSU band present the Krebs cycle: http://www.youtube.com/watch?v=FgXnH087JIk&feature=related.

Results of the Krebs Cycle

After the second turn through the Krebs cycle, the original glucose molecule has been broken down completely. All six of its carbon atoms have combined with oxygen to form carbon dioxide. The energy from its chemical bonds has been stored in a total of 16 energy-carrier molecules. These molecules are:

- 4 ATP (including 2 from glycolysis)
- 10 NADH (including 2 from glycolysis)
- 2 $FADH_2$

The Krebs cycle is reviewed at http://www.youtube.com/user/khanacademy#p/c/7A9646BC5110CF64/23/juM2ROSLWfw.



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Cellular Respiration Stage III: Electron Transport

Electron transport is the final stage of aerobic respiration. In this stage, energy from NADH and $FADH_2$, which result from the Krebs cycle, is transferred to ATP. Can you predict how this happens? (*Hint:* How does electron transport occur in photosynthesis?)

See http://www.youtube.com/watch?v=1engJR_XWVU&feature=related for an overview of the electron transport chain.

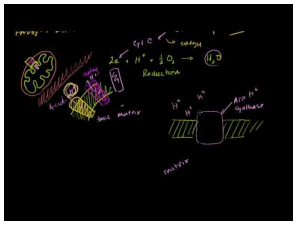
Transporting Electrons

High-energy electrons are released from NADH and $FADH_2$, and they move along electron transport chains, like those used in photosynthesis. The electron transport chains are on the inner membrane of the mitochondrion. As the high-energy electrons are transported along the chains, some of their energy is captured. This energy is used to pump hydrogen ions (from NADH and $FADH_2$) across the inner membrane, from the matrix into the intermembrane space. Electron transport in a mitochondrion is shown in **Figure** 4.14. You can also see an animation of the process at this link: http://www.youtube.com/watch?v=Idy2XAlZIVA&feature=related.

Making ATP

The pumping of hydrogen ions across the inner membrane creates a greater concentration of the ions in the intermembrane space than in the matrix. This chemiosmotic gradient causes the ions to flow back across the membrane into the matrix, where their concentration is lower. ATP synthase acts as a channel protein, helping the hydrogen ions cross the membrane. It also acts as an enzyme, forming ATP from ADP and inorganic phosphate. After passing through the electron-transport chain, the "spent" electrons combine with oxygen to form water. This is why oxygen is needed; in the absence of oxygen, this process cannot occur. You can see how all these events occur at the following link: http://www.sp.uconn.edu/ terry/images/anim/ATPmito.html.

A summary of this process can be seen at the following sites: http://www.youtube.com/user/khanacademy#p/c/7A9646BC5110CF64/24/mfgCcFXUZRk (17:16) and http://www.youtube.com/user/khanacademy#p/c/7A9646BC5110CF64/25/W_Q17tqw_7A (4:59).



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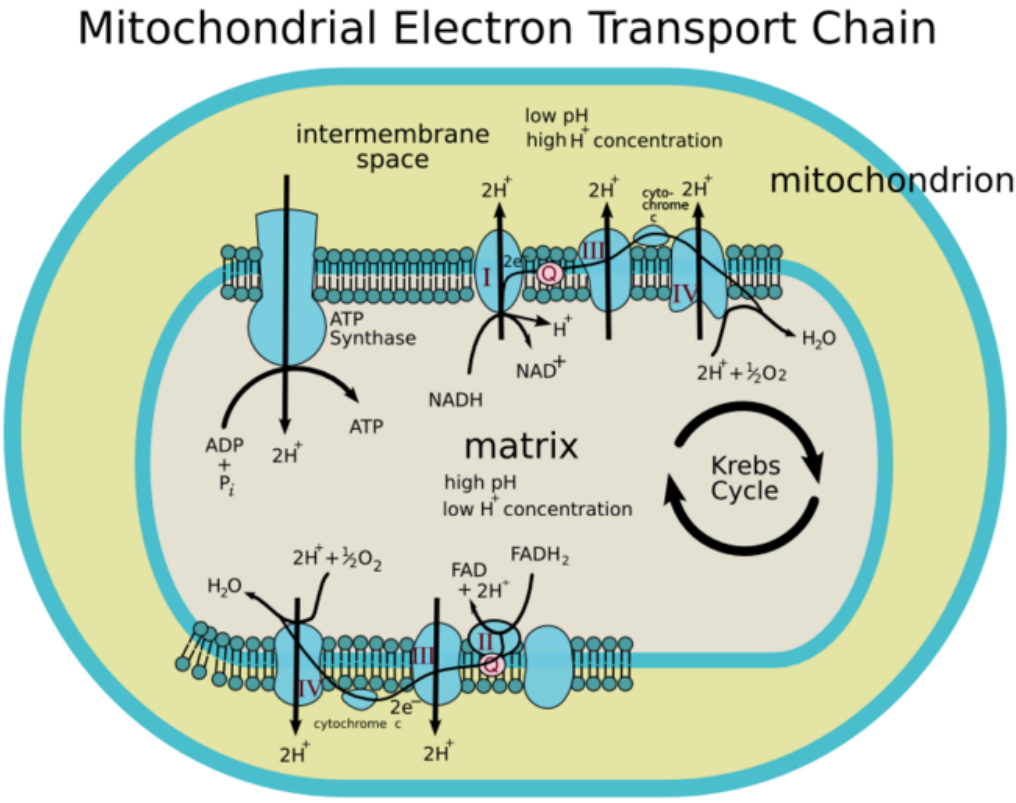
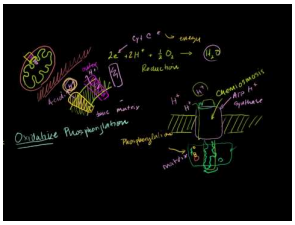


FIGURE 4.14

Electron-transport chains on the inner membrane of the mitochondrion carry out the last stage of cellular respiration.



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How Much ATP?

You have seen how the three stages of aerobic respiration use the energy in glucose to make ATP. How much ATP is produced in all three stages? Glycolysis produces 2 ATP molecules, and the Krebs cycle produces 2 more. Electron transport begins with several molecules of NADH and $FADH_2$ from the Krebs cycle and transfers their energy into as many as 34 more ATP molecules. All told, then, up to 38 molecules of ATP can be produced from just one molecule of glucose in the process of aerobic respiration.

Lesson Summary

- Cellular respiration uses energy in glucose to make ATP. Aerobic ("oxygen-using") respiration occurs in three stages: glycolysis, the Krebs cycle, and electron transport.
- In glycolysis, glucose is split into two molecules of pyruvate. This results in a net gain of two ATP molecules.
- Life first evolved in the absence of oxygen, and glycolysis does not require oxygen. Therefore, glycolysis was probably the earliest way of making ATP from glucose.
- The Krebs cycle and electron transport occur in the mitochondria. The Krebs cycle takes place in the matrix, and electron transport takes place on the inner membrane.
- During the Krebs cycle, pyruvate undergoes a series of reactions to produce two more molecules of ATP and also several molecules of NADH and $FADH_2$.
- During electron transport, energy from NADH and $FADH_2$ is used to make many more molecules of ATP.
- In all three stages of aerobic respiration, up to 38 molecules of ATP may be produced from a single molecule of glucose.

Lesson Review Questions

Recall

1. List the stages of aerobic respiration in the order in which they occur.

2. Describe what happens during glycolysis. How many ATP molecules are gained during this stage?

3. Define aerobic and anaerobic respiration.

4. What role do mitochondria play in cellular respiration?

5. What are the products of the Krebs cycle?

6. What is the maximum number of ATP molecules that can be produced during the electron transport stage of aerobic respiration?

Apply Concepts

7. When you exhale onto a cold window pane, water vapor in your breath condenses on the glass. Where does the water vapor come from?

8. Assume that a new species of organism has been discovered. Scientists have observed its cells under a microscope and determined that they lack mitochondria. What type of cellular respiration would you predict that the new species uses? Explain your prediction.

Think Critically

9. Why do scientists think that glycolysis evolved before the other stages of cellular respiration?

10. Explain why two turns of the Krebs cycle are needed for each molecule of glucose.

Points to Consider

The last two stages of aerobic respiration require oxygen. However, not all organisms live in places where there is a plentiful supply of oxygen.

- How do you think organisms get energy from glucose to make ATP if they cannot use oxygen?
- Do they just use glycolysis, which produces only two ATP molecules? Or do you think there might be other steps involved?

4.4 Anaerobic Respiration

Lesson Objectives

- Define fermentation.
- Describe lactic acid fermentation and alcoholic fermentation.
- Compare the advantages of aerobic and anaerobic respiration.

Vocabulary

alcoholic fermentation type of anaerobic respiration that includes glycolysis followed by the conversion of pyruvic acid to ethanol and carbon dioxide and the formation of NAD^+

fermentation type of anaerobic respiration that includes glycolysis followed by the conversion of pyruvic acid to one or more other compounds and the formation of NAD^+

lactic acid fermentation type of anaerobic respiration that includes glycolysis followed by the conversion of pyruvic acid to lactic acid and the formation of NAD^+

Introduction

Today, most living things use oxygen to make ATP from glucose. However, many living things can also make ATP without oxygen. This is true of some plants and fungi and also of many bacteria. These organisms use aerobic respiration when oxygen is present, but when oxygen is in short supply, they use anaerobic respiration instead. Certain bacteria can only use anaerobic respiration. In fact, they may not be able to survive at all in the presence of oxygen.

Fermentation

An important way of making ATP without oxygen is called **fermentation**. It involves glycolysis but not the other two stages of aerobic respiration. Many bacteria and yeasts carry out fermentation. People use these organisms to make yogurt, bread, wine, and biofuels. Human muscle cells also use fermentation. This occurs when muscle cells cannot get oxygen fast enough to meet their energy needs through aerobic respiration. There are two types of fermentation: lactic acid fermentation and alcoholic fermentation. Both types of fermentation are described below. You can also watch animations of both types at this link: http://www.cst.cmich.edu/users/schul1te/animations/fermentation.swf.

Lactic Acid Fermentation

In **lactic acid fermentation**, pyruvic acid from glycolysis changes to lactic acid. This is shown in **Figure** 4.15. In the process, NAD^+ forms from NADH. NAD^+, in turn, lets glycolysis continue. This results in additional molecules of ATP. This type of fermentation is carried out by the bacteria in yogurt. It is also used by your own muscle cells when you work them hard and fast.

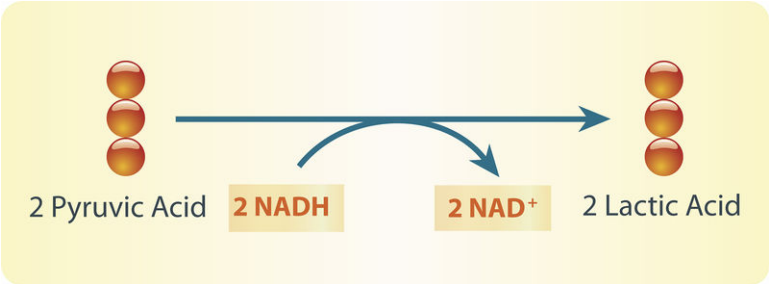

FIGURE 4.15

Lactic acid fermentation produces lactic acid and NAD+. The NAD+ cycles back to allow glycolysis to continue so more ATP is made. Each circle represents a carbon atom.

Did you ever run a race and notice that your muscles feel tired and sore afterward? This is because your muscle cells used lactic acid fermentation for energy. This causes lactic acid to build up in the muscles. It is the buildup of lactic acid that makes the muscles feel tired and sore.

Alcoholic Fermentation

In **alcoholic fermentation**, pyruvic acid changes to alcohol and carbon dioxide. This is shown in **Figure** 4.16. NAD^+ also forms from NADH, allowing glycolysis to continue making ATP. This type of fermentation is carried out by yeasts and some bacteria. It is used to make bread, wine, and biofuels.

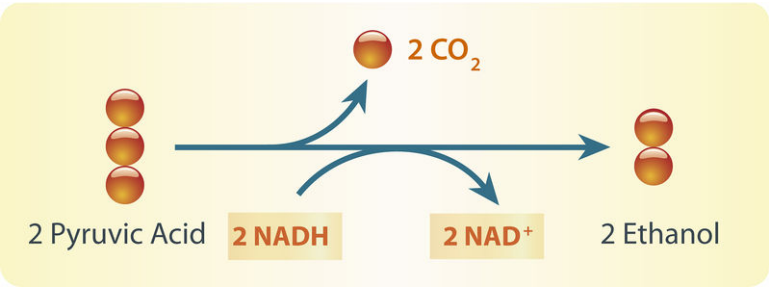

FIGURE 4.16

Alcoholic fermentation produces ethanol and NAD+. The NAD+ allows glycolysis to continue making ATP.

Have your parents ever put corn in the gas tank of their car? They did if they used gas containing ethanol. Ethanol is produced by alcoholic fermentation of the glucose in corn or other plants. This type of fermentation also explains why bread dough rises. Yeasts in bread dough use alcoholic fermentation and produce carbon dioxide gas. The gas forms bubbles in the dough, which cause the dough to expand. The bubbles also leave small holes in the bread after it bakes, making the bread light and fluffy. Do you see the small holes in the slice of bread in **Figure** 4.17?

FIGURE 4.17

The small holes in bread are formed by bubbles of carbon dioxide gas. The gas was produced by alcoholic fermentation carried out by yeast.

Aerobic vs. Anaerobic Respiration: A Comparison

Aerobic respiration evolved after oxygen was added to Earth's atmosphere. This type of respiration is useful today because the atmosphere is now 21% oxygen. However, some anaerobic organisms that evolved before the atmosphere contained oxygen have survived to the present. Therefore, anaerobic respiration must also have advantages.

Advantages of Aerobic Respiration

A major advantage of aerobic respiration is the amount of energy it releases. Without oxygen, organisms can just split glucose into two molecules of pyruvate. This releases only enough energy to make two ATP molecules. With oxygen, organisms can break down glucose all the way to carbon dioxide. This releases enough energy to produce up to 38 ATP molecules. Thus, aerobic respiration releases much more energy than anaerobic respiration. The amount of energy produced by aerobic respiration may explain why aerobic organisms came to dominate life on Earth. It may also explain how organisms were able to become multicellular and increase in size.

Advantages of Anaerobic Respiration

One advantage of anaerobic respiration is obvious. It lets organisms live in places where there is little or no oxygen. Such places include deep water, soil, and the digestive tracts of animals such as humans (see **Figure** 4.18).

Another advantage of anaerobic respiration is its speed. It produces ATP very quickly. For example, it lets your muscles get the energy they need for short bursts of intense activity (see **Figure** 4.19). Aerobic respiration, on the other hand, produces ATP more slowly.

Lesson Summary

- Fermentation is a way of making ATP from glucose without oxygen. There are two types of fermentation: lactic acid fermentation and alcoholic fermentation.
- Lactic acid fermentation changes pyruvic acid to lactic acid and forms NAD^+. The NAD^+ allows glycolysis to continue so it can make more ATP.
- Alcohol fermentation changes pyruvic acid to ethanol and carbon dioxide and forms NAD^+. Again, the NAD^+ allows glycolysis to keep making ATP.

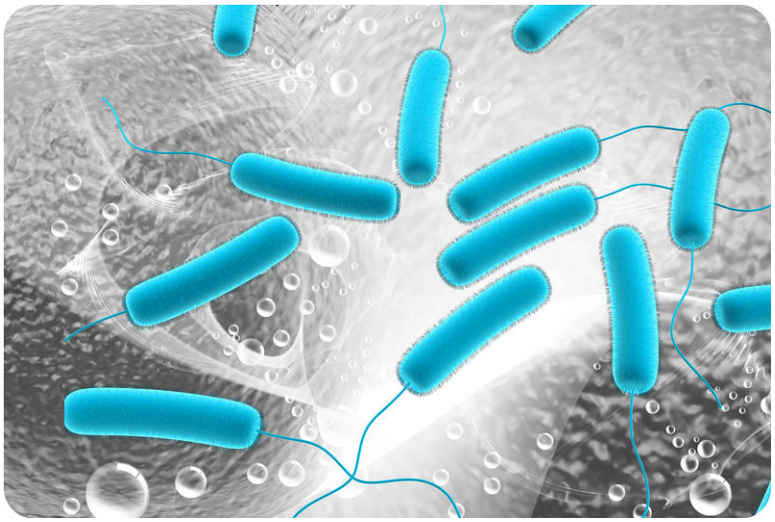

FIGURE 4.18

E. coli bacteria are anaerobic bacteria that live in the human digestive tract.

FIGURE 4.19

The muscles of these hurdlers need to use anaerobic respiration for energy. It gives them the energy they need for the short-term, intense activity of this sport.

- Aerobic respiration produces much more ATP than anaerobic respiration. However, anaerobic respiration occurs more quickly.

Lesson Review Questions

Recall

1. What is fermentation?

2. Name two types of fermentation.

3. What is the main advantage of aerobic respiration? Of anaerobic respiration?

4. What process produces fuel for motor vehicles from living plant products? What is the waste product of this process?

Apply Concepts

5. Tanya is on the high school track team and runs the 100-meter sprint. Marissa is on the cross-country team and runs 5-kilometer races. Explain which type of respiration the muscle cells in each runner's legs use.

Think Critically

6. Compare and contrast lactic acid fermentation and alcoholic fermentation. Include examples of organisms that use each type of fermentation.

7. Explain why bread dough rises when it is set aside in a warm place.

Points to Consider

Two important functions of cells are making food and using it for energy. Photosynthesis and cellular respiration are the processes that carry out these functions. Other important functions of cells are growing and dividing.

- Do you know how cells grow? What do you think controls the growth of cells?
- How do you think cells divide? Do all cells divide in the same way?

4.5 References

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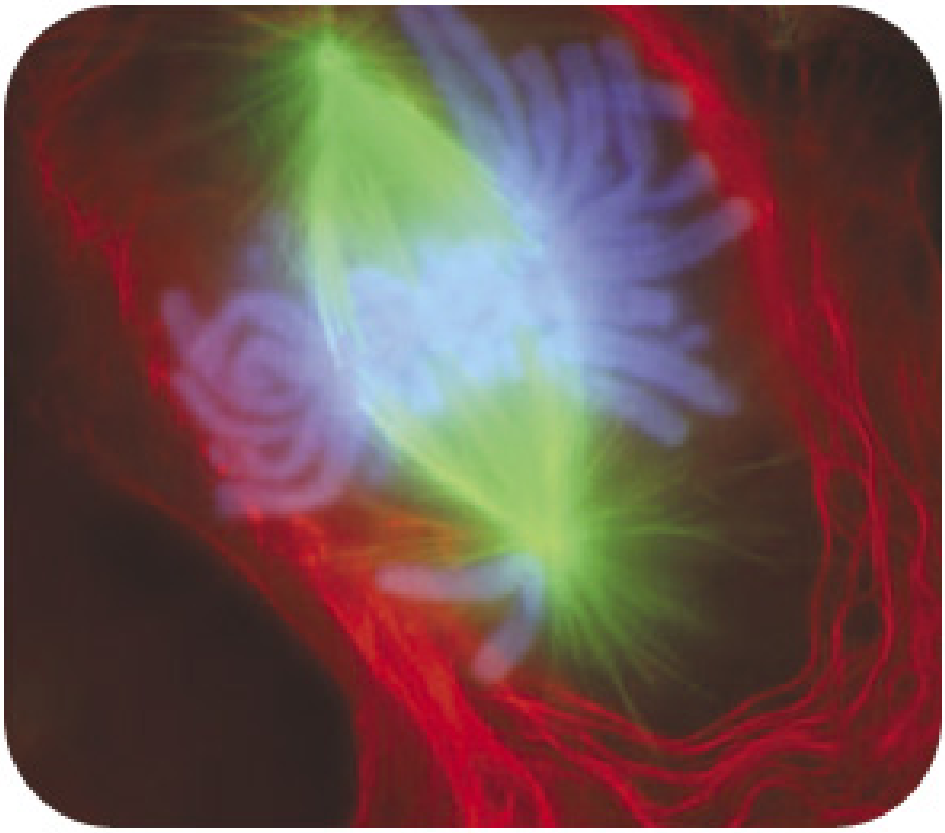
CHAPTER 5 The Cell Cycle, Mitosis, and Meiosis

Chapter Outline



What do you think this colorful picture shows? If you guessed that it's a picture of a cell undergoing cell division, you are right. In fact, the picture is an image of a lung cell stained with fluorescent dyes undergoing mitosis, specifically during early anaphase. You will read about mitosis, a type of cell division, in this chapter.

Cell division is just one of the stages that all cells go through during their life. This includes cells that are harmful, such as cancer cells. Cancer cells divide more often than normal cells, and grow out of control. In fact, this is how cancer cells cause illness. In this chapter, you will read about how cells divide, what other stages cells go through, and what causes cancer cells to divide out of control and harm the body.

5.1 Cell Division and the Cell Cycle

Lesson Objectives

- Contrast cell division in prokaryotes and eukaryotes.
- Identify the phases of the eukaryotic cell cycle.
- Explain how the cell cycle is controlled.
- Define cancer, and relate it to the cell cycle.

Vocabulary

binary fission type of cell division that occurs in prokaryotic cells in which a parent cells divides into two identical daughter cells

cancer disease that occurs when the cell cycle is no longer regulated and cells divide out of control

cell cycle repeating series of events that a cell goes through during its life, including growth, DNA, synthesis, and cell division

cell division process in which a parent cell divides to form two daughter cells

cytokinesis splitting of the cytoplasm to form daughter cells when a cell divides

DNA replication process of copying of DNA prior to cell division

interphase stage of the eukaryotic cell cycle when the cell grows, synthesizes DNA, and prepares to divide

mitosis process in which the nucleus of a eukaryotic cell divides

tumor abnormal mass of cells that may be cancerous

Introduction

You consist of a great many cells, but like all other organisms, you started life as a single cell. How did you develop from a single cell into an organism with trillions of cells? The answer is cell division. After cells grow to their maximum size, they divide into two new cells. These new cells are small at first, but they grow quickly and eventually divide and produce more new cells. This process keeps repeating in a continuous cycle.

Cell Division

Cell division is the process in which one cell, called the parent cell, divides to form two new cells, referred to as daughter cells. How this happens depends on whether the cell is prokaryotic or eukaryotic.

Cell division is simpler in prokaryotes than eukaryotes because prokaryotic cells themselves are simpler. Prokaryotic cells have a single circular chromosome, no nucleus, and few other organelles. Eukaryotic cells, in contrast, have multiple chromosomes contained within a nucleus and many other organelles. All of these cell parts must be duplicated and then separated when the cell divides.

Cell Division in Prokaryotes

Most prokaryotic cells divide by the process of **binary fission**. A bacterial cell dividing this way is depicted in **Figure** 5.1. You can also watch an animation of binary fission at this link: http://en.wikipedia.org/wiki/File:Binary_fission_anim.gif.

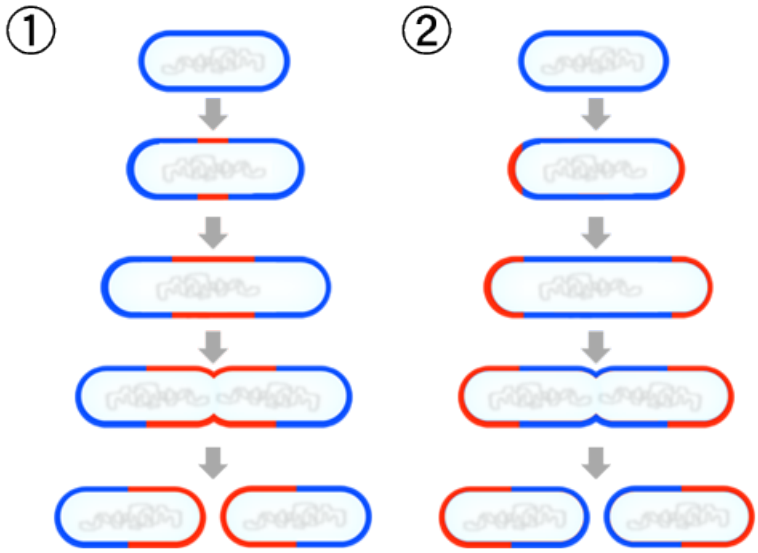


FIGURE 5.1

Binary Fission in a Bacterial Cell. Cell division is relatively simple in prokaryotic cells. The two cells are dividing by binary fission. Blue and red lines indicate old and newly-generated bacterial cell walls, respectively. Eventually the parent cell will pinch apart to form two identical daughter cells. Left, growth at the center of bacterial body. Right, apical growth from the ends of the bacterial body.

Binary fission can be broken down into a series of three steps, although it is actually a continuous process. The steps are described below and also illustrated in **Figure** 5.2. They include DNA replication, chromosome segregation, and cytokinesis.

- **Step 1: DNA Replication**. Just before the cell divides, its DNA is copied in a process called **DNA replication**. This results in two identical chromosomes instead of just one. This step is necessary so that when the cell divides, each daughter cell will have its own chromosome.
- **Step 2: Chromosome Segregation**. The two chromosomes segregate, or separate, and move to opposite ends (known as *poles*) of the cell.
- **Step 3: Cytokinesis**. A new plasma membrane starts growing into the center of the cell, and the cytoplasm splits apart, forming two daughter cells. This process is called **cytokinesis**. The two daughter cells that result are genetically identical to each other and to the parent cell.

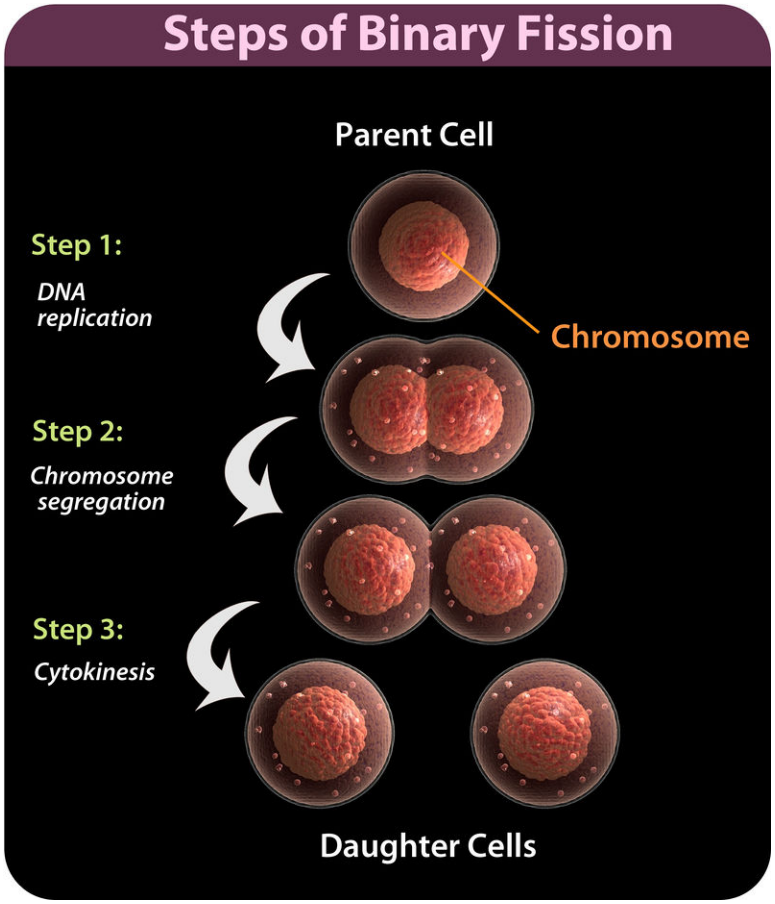


FIGURE 5.2

Steps of Binary Fission. Prokaryotic cells divide by binary fission. This is also how many single-celled organisms reproduce.

Cell Division in Eukaryotes

Cell division is more complex in eukaryotes than prokaryotes. Prior to dividing, all the DNA in a eukaryotic cell's multiple chromosomes is replicated. Its organelles are also duplicated. Then, when the cell divides, it occurs in two major steps:

- The first step is **mitosis**, a multi-phase process in which the nucleus of the cell divides. During mitosis, the nuclear membrane breaks down and later reforms. The chromosomes are also sorted and separated to ensure that each daughter cell receives a complete set of chromosomes. Mitosis is described in greater detail in Lesson 5.2.
- The second major step is cytokinesis. As in prokaryotic cells, during this step the cytoplasm divides and two daughter cells form.

The Cell Cycle

Cell division is just one of several stages that a cell goes through during its lifetime. The **cell cycle** is a repeating series of events that include growth, DNA synthesis, and cell division. The cell cycle in prokaryotes is quite simple: the cell grows, its DNA replicates, and the cell divides. In eukaryotes, the cell cycle is more complicated.

Eukaryotic Cell Cycle

The diagram in **Figure** 5.3 represents the cell cycle of a eukaryotic cell. As you can see, the eukaryotic cell cycle has several phases. The mitosis phase (M) actually includes both mitosis and cytokinesis. This is when the nucleus and then the cytoplasm divide. The other three phases (G1, S, and G2) are generally grouped together as **interphase**. During interphase, the cell grows, performs routine life processes, and prepares to divide. These phases are discussed below. You can watch a eukaryotic cell going through these phases of the cell cycle at the following link: http://www.cellsalive.com/cell_cycle.htm.

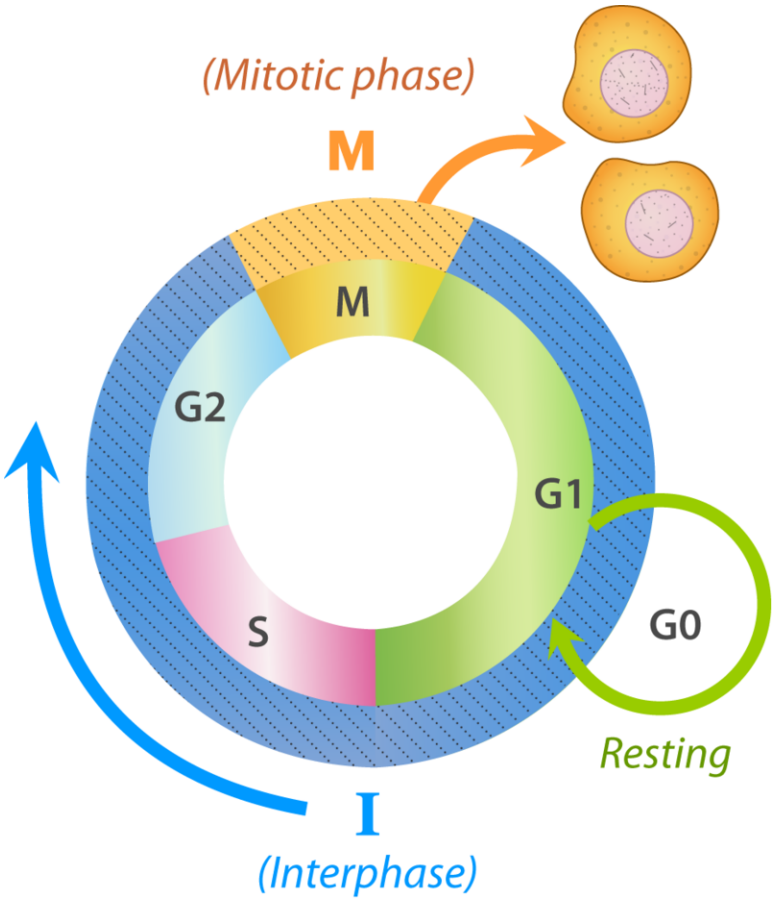


FIGURE 5.3

Eukaryotic Cell Cycle. This diagram represents the cell cycle in eukaryotes. The G1, S, and G2 phases make up interphase (I). The M phase includes mitosis and cytokinesis. After the M phase, two cells result.

Interphase

Interphase of the eukaryotic cell cycle can be subdivided into the following three phases, which are represented in **Figure** 5.3:

- **Growth Phase 1 (G1):** during this phase, the cell grows rapidly, while performing routine metabolic processes. It also makes proteins needed for DNA replication and copies some of its organelles in preparation for cell division. A cell typically spends most of its life in this phase.
- **Synthesis Phase (S):** during this phase, the cell's DNA is copied in the process of DNA replication.
- **Growth Phase 2 (G2):** during this phase, the cell makes final preparations to divide. For example, it makes additional proteins and organelles.

Control of the Cell Cycle

If the cell cycle occurred without regulation, cells might go from one phase to the next before they were ready. What controls the cell cycle? How does the cell know when to grow, synthesize DNA, and divide? The cell cycle is controlled mainly by regulatory proteins. These proteins control the cycle by signaling the cell to either start or delay the next phase of the cycle. They ensure that the cell completes the previous phase before moving on. Regulatory proteins control the cell cycle at key checkpoints, which are shown in **Figure** 5.4. There are a number of main checkpoints.

- The G1 checkpoint, just before entry into S phase, makes the key decision of whether the cell should divide.
- The S checkpoint determines if the DNA has been replicated properly.
- The mitotic spindle checkpoint occurs at the point in metaphase where all the chromosomes should have aligned at the mitotic plate.

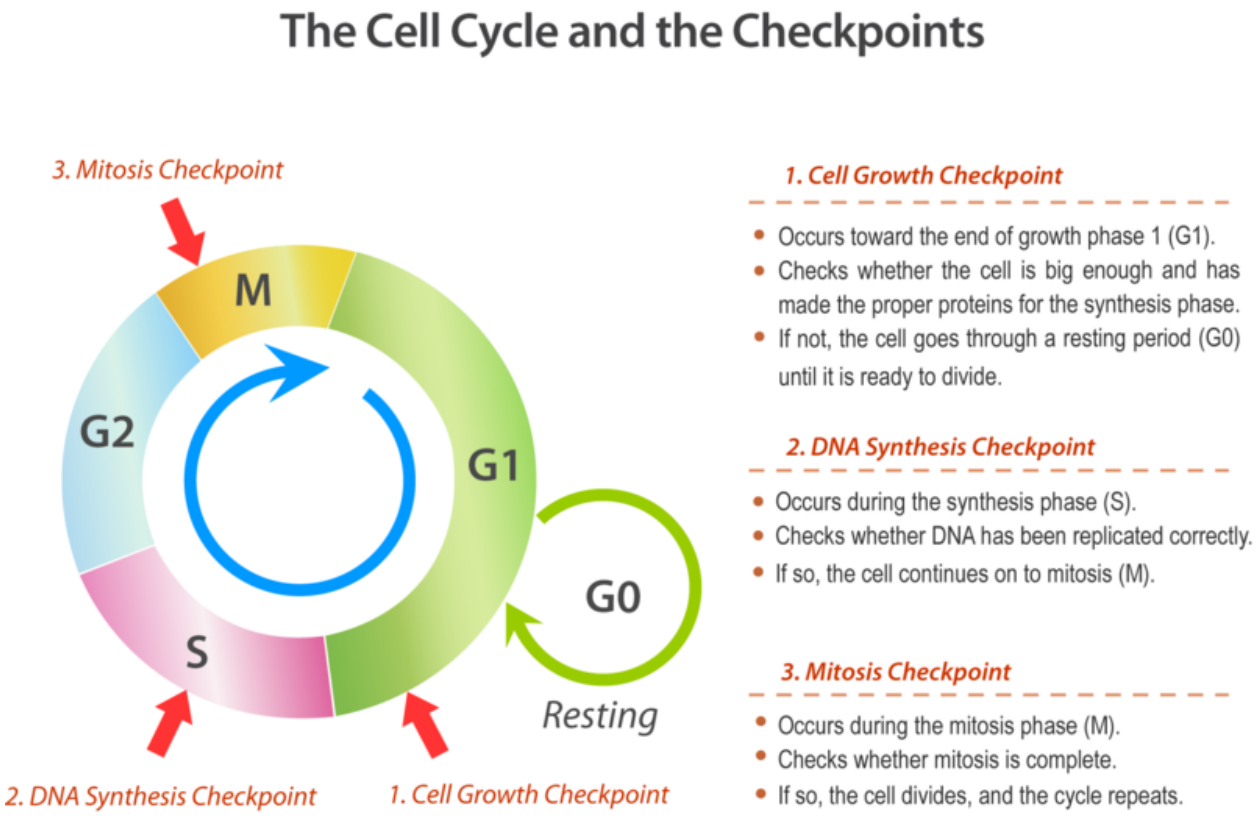


FIGURE 5.4

Checkpoints (arrows) in the eukaryotic cell cycle ensure that the cell is ready to proceed before it moves on to the next phase of the cycle.

Cancer and the Cell Cycle

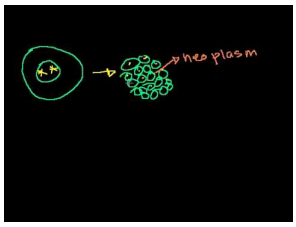
Cancer is a disease that occurs when the cell cycle is no longer regulated. This may happen because a cell's DNA becomes damaged. Damage can occur due to exposure to hazards such as radiation or toxic chemicals. Cancerous

cells generally divide much faster than normal cells. They may form a mass of abnormal cells called a **tumor** (see **Figure** 5.5). The rapidly dividing cells take up nutrients and space that normal cells need. This can damage tissues and organs and eventually lead to death.

FIGURE 5.5

These cells are cancer cells, growing out of control and forming a tumor.

Cancer is discussed in the video at http://www.youtube.com/user/khanacademy#p/c/7A9646BC5110CF64/11/RZhL7LDPk8w.



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Lesson Summary

- Cell division is part of the life cycle of virtually all cells. It is a more complicated process in eukaryotic than prokaryotic cells because eukaryotic cells have multiple chromosomes and a nucleus.
- The cell cycle is a repeating series of events that cells go through. It includes growth, DNA synthesis, and cell division. In eukaryotic cells, there are two growth phases, and cell division includes mitosis.
- The cell cycle is controlled by regulatory proteins at three key checkpoints in the cycle. The proteins signal the cell to either start or delay the next phase of the cycle.
- Cancer is a disease that occurs when the cell cycle is no longer regulated. Cancer cells grow rapidly and may form a mass of abnormal cells called a tumor.

Lesson Review Questions

Recall

1. Describe binary fission.
2. What is mitosis?
3. Identify the phases of the eukaryotic cell cycle.
4. What happens during interphase?
5. Define cancer.

Apply Concepts

6. How might the relationship between cancer and the cell cycle be used in the search for causes of cancer?

Think Critically

7. Cells go through a series of events that include growth, DNA synthesis, and cell division. Why are these events best represented by a cycle diagram?
8. Contrast cell division in prokaryotes and eukaryotes. Why are the two types of cell division different?
9. Explain how the cell cycle is regulated.
10. Why is DNA replication essential to the cell cycle?

Points to Consider

When a eukaryotic cell divides, the nucleus divides first in the process of mitosis.

- What do you think happens during mitosis? Can you predict what molecules and cell structures are involved in this process?
- How do you think mitosis might differ from binary fission? What steps might be involved in mitosis?

5.2 Chromosomes and Mitosis

Lesson Objectives

- Describe chromosomes and their role in mitosis.
- Outline the phases of mitosis.

Vocabulary

anaphase third phase of mitosis during which sister chromatids separate and move to opposite poles of the cell

centromere region of sister chromatids where they are joined together

chromatid one of two identical copies of a chromosome that are joined together at a centromere before a cell divides

chromatin grainy material that DNA forms when it is not coiled into chromosomes

chromosome coiled structure made of DNA and proteins containing sister chromatids that is the form in which the genetic material of a cell goes through cell division

gene unit of DNA on a chromosome that is encoded with the instructions for a single protein

homologous chromosomes pair of chromosomes that have the same size and shape and contain the same genes

metaphase second phase of mitosis during which chromosomes line up at the equator of the cell

prophase first phase of mitosis during which chromatin condense into chromosomes, the nuclear envelope breaks down, centrioles separate, and a spindle begins to form

telophase last stage of mitosis during which chromosomes uncoil to form chromatin, the spindle breaks down, and new nuclear membranes form

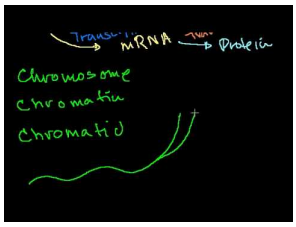
Introduction

In eukaryotic cells, the nucleus divides before the cell itself divides. The process in which the nucleus divides is called mitosis. Before mitosis occurs, a cell's DNA is replicated. This is necessary so that each daughter cell will have a complete copy of the genetic material from the parent cell. How is the replicated DNA sorted and separated so that each daughter cell gets a complete set of the genetic material? To understand how this happens, you need to know more chromosomes.

Chromosomes

Chromosomes are coiled structures made of DNA and proteins. Chromosomes are the form of the genetic material of a cell during cell division. During other phases of the cell cycle, DNA is not coiled into chromosomes. Instead, it exists as a grainy material called **chromatin**.

The vocabulary of DNA: chromosomes, chromatids, chromatin, transcription, translation, and replication is discussed at http://www.youtube.com/user/khanacademy#p/c/7A9646BC5110CF64/6/s9HPNwXd9fk (18:23).



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Chromatids and the Centromere

DNA condenses and coils into the familiar X-shaped form of a chromosome, shown in **Figure** 5.6, only after it has replicated. (You can watch DNA coiling into a chromosome at the link below.) Because DNA has already replicated, each chromosome actually consists of two identical copies. The two copies are called sister **chromatids**. They are attached to one another at a region called the **centromere**. A remarkable animation can be viewed at http://www.hhmi.org/biointeractive/media/DNAi_packaging_vo2-sm.mov.

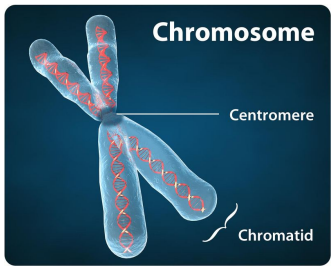


FIGURE 5.6

Chromosome. After DNA replicates, it forms chromosomes like the one shown here.

Chromosomes and Genes

The DNA of a chromosome is encoded with genetic instructions for making proteins. These instructions are organized into units called **genes**. Most genes contain the instructions for a single protein. There may be hundreds or even thousands of genes on a single chromosome.

Human Chromosomes

Human cells normally have two sets of chromosomes, one set inherited from each parent. There are 23 chromosomes in each set, for a total of 46 chromosomes per cell. Each chromosome in one set is matched by a chromosome of the same type in the other set, so there are actually 23 pairs of chromosomes per cell. Each pair consists of chromosomes of the same size and shape that also contain the same genes. The chromosomes in a pair are known as **homologous chromosomes**.

Mitosis and Cytokinesis

During mitosis, when the nucleus divides, the two chromatids that make up each chromosome separate from each other and move to opposite poles of the cell. This is shown in **Figure** 5.7. You can watch an animation of the process at the following link: http://www.biology.arizona.edu/Cell_bio/tutorials/cell_cycle/MitosisFlash.html.

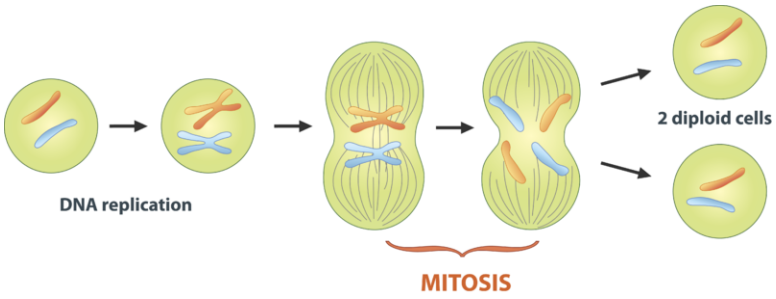


FIGURE 5.7

Mitosis is the phase of the eukaryotic cell cycle that occurs between DNA replication and the formation of two daughter cells. What happens during mitosis?

Mitosis actually occurs in four phases. The phases are called prophase, metaphase, anaphase, and telophase. They are shown in **Figure** 5.8 and described in greater detail in the following sections.

Prophase

The first and longest phase of mitosis is **prophase**. During prophase, chromatin condenses into chromosomes, and the nuclear envelope, or membrane, breaks down. In animal cells, the centrioles near the nucleus begin to separate and move to opposite poles of the cell. As the centrioles move, a spindle starts to form between them. The spindle, shown in **Figure** 5.9, consists of fibers made of microtubules.

Metaphase

During **metaphase**, spindle fibers attach to the centromere of each pair of sister chromatids (see **Figure** 5.10). The sister chromatids line up at the equator, or center, of the cell. The spindle fibers ensure that sister chromatids will separate and go to different daughter cells when the cell divides.

Anaphase

During **anaphase**, sister chromatids separate and the centromeres divide. The sister chromatids are pulled apart by the shortening of the spindle fibers. This is like reeling in a fish by shortening the fishing line. One sister chromatid moves to one pole of the cell, and the other sister chromatid moves to the opposite pole. At the end of anaphase, each pole of the cell has a complete set of chromosomes.

Telophase

During **telophase**, the chromosomes begin to uncoil and form chromatin. This prepares the genetic material for directing the metabolic activities of the new cells. The spindle also breaks down, and new nuclear membranes form.

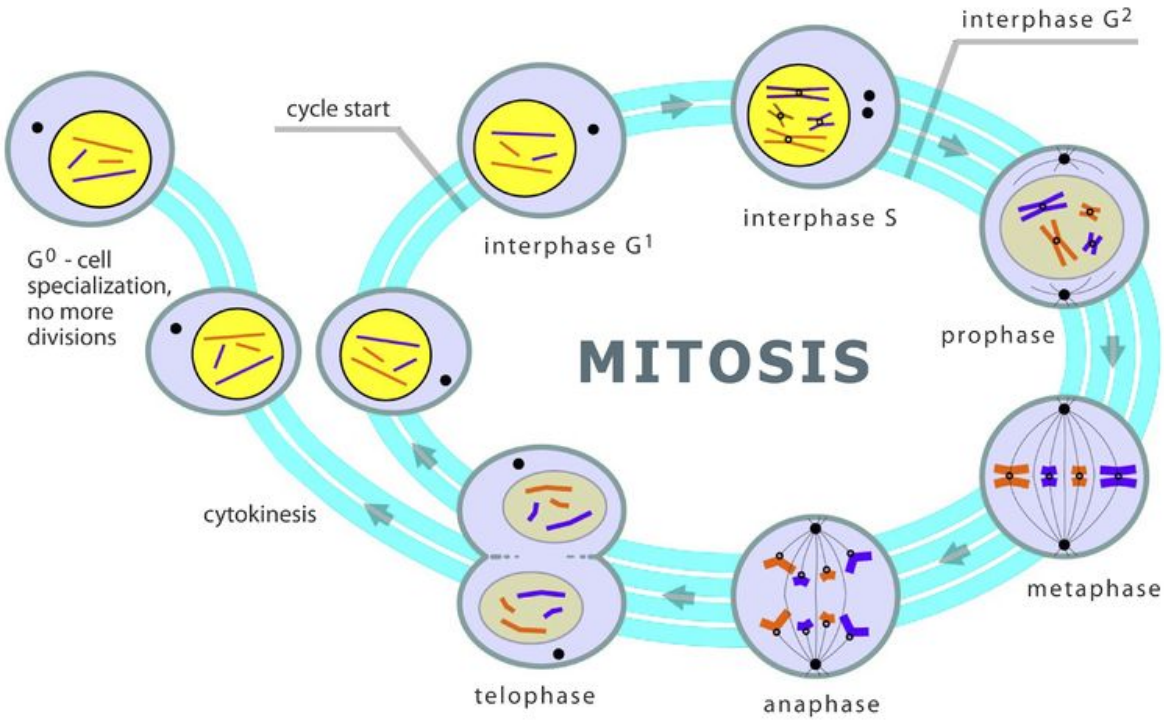


FIGURE 5.8

Mitosis in the Eukaryotic Cell Cycle. Mitosis is the multi-phase process in which the nucleus of a eukaryotic cell divides.

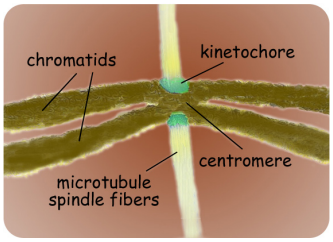


FIGURE 5.9

Spindle. The spindle starts to form during prophase of mitosis. Kinetochores on the spindle attach to the centromeres of sister chromatids.

Cytokinesis

Cytokinesis is the final stage of cell division in eukaryotes as well as prokaryotes. During cytokinesis, the cytoplasm splits in two and the cell divides. Cytokinesis occurs somewhat differently in plant and animal cells, as shown in **Figure** 5.11. In animal cells, the plasma membrane of the parent cell pinches inward along the cell's equator until two daughter cells form. In plant cells, a cell plate forms along the equator of the parent cell. Then, a new plasma

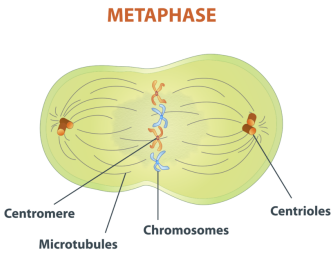


FIGURE 5.10

Chromosomes, consisting of sister chromatids, line up at the equator of the cell during metaphase.

membrane and cell wall form along each side of the cell plate.

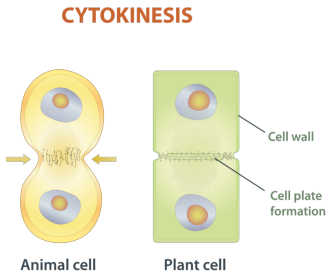
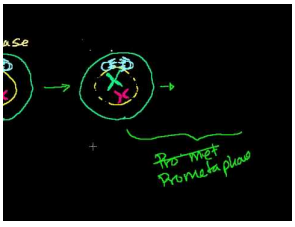


FIGURE 5.11

Cytokinesis is the final stage of eukaryotic cell division. It occurs differently in animal and plant cells.

The phases of mitosis are discussed in the video: http://www.youtube.com/user/khanacademy#p/c/7A9646BC5110CF64/8/LLKX_4DHE3I.



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Lesson Summary

- Chromosomes are coiled structures made of DNA and proteins. They form after DNA replicates and are the form in which the genetic material goes through cell division. Chromosomes contain genes, which code for proteins.
- Cell division in eukaryotic cells includes mitosis, in which the nucleus divides, and cytokinesis, in which the cytoplasm divides and daughter cells form.
- Mitosis occurs in four phases, called prophase, metaphase, anaphase, and telophase.

Lesson Review Questions

Recall

1. What are chromosomes? When do they form?
2. Identify the chromatids and the centromere of a chromosome.
3. List the phases of mitosis.
4. What happens during prophase of mitosis?
5. During which phase of mitosis do sister chromatids separate?
6. Describe what happens during cytokinesis in animal cells.

Apply Concepts

7. If a cell skipped metaphase during mitosis, how might this affect the two daughter cells?

Think Critically

8. Explain how chromosomes are related to chromatin. Why are chromosomes important for mitosis?
9. Explain the significance of the spindle in mitosis.

Points to Consider

Cell division occurs not only as organisms grow. It also occurs when they reproduce.

- What role do you think cell division plays when prokaryotes such as bacteria reproduce?
- How do you think cell division is involved in the reproduction of eukaryotes such as humans?

5.3 Reproduction and Meiosis

Lesson Objectives

- Compare and contrast asexual and sexual reproduction.
- Give an overview of sexual reproduction, and outline the phases of meiosis.
- Explain why sexual reproduction leads to variation in offspring.
- Define life cycle, and identify different types of sexual life cycles.

Vocabulary

asexual reproduction reproduction that involves a single parent and results in offspring that are all genetically identical to the parent

crossing-over exchange of genetic material between homologous chromosomes when they are closely paired during meiosis I

diploid having two of each type of chromosome

egg female gamete

fertilization union of two gametes that produces a diploid zygote

gamete reproductive cell produced during meiosis that has the haploid number of chromosomes

gametogenesis development of haploid cells into gametes such as sperm and egg

haploid having only one chromosome of each type

independent assortment independent segregation of chromosomes to gametes during meiosis

life cycle series of stages a sexually reproducing organism goes through from one generation to the next

meiosis type of cell division in which the number of chromosomes is reduced by half and four haploid cells result

sexual reproduction type of reproduction that involves the fertilization of gametes produced by two parents and produces genetically variable offspring

sperm male gamete

zygote diploid cell that forms when two haploid gametes unite during fertilization

Introduction

Cell division is how organisms grow and repair themselves. It is also how they produce offspring. Many single-celled organisms reproduce by binary fission. The parent cell simply divides to form two daughter cells that are identical to the parent. In many other organisms, two parents are involved, and the offspring are not identical to the parents. In fact, each offspring is unique. Look at the family in **Figure** 5.12. The children resemble their parents, but they are not identical to them. Instead, each has a unique combination of characteristics inherited from both parents. In this lesson, you will learn how this happens.

FIGURE 5.12

Family Portrait: Mother, Daughter, Father, and Son. Children resemble their parents, but they are never identical to them. Do you know why this is the case?

Reproduction: Asexual vs. Sexual

Reproduction is the process by which organisms give rise to offspring. It is one of the defining characteristics of living things. There are two basic types of reproduction: asexual reproduction and sexual reproduction.

Asexual Reproduction

Asexual reproduction involves a single parent. It results in offspring that are genetically identical to each other and to the parent. All prokaryotes and some eukaryotes reproduce this way. There are several different methods of asexual reproduction. They include binary fission, fragmentation, and budding.

- Binary fission occurs when a parent cell splits into two identical daughter cells of the same size. This process was described in detail in Lesson 5.1.
- Fragmentation occurs when a parent organism breaks into fragments, or pieces, and each fragment develops into a new organism. Starfish, like the one in **Figure** 5.13, reproduce this way. A new starfish can develop from a single ray, or arm.
- Budding occurs when a parent cell forms a bubble-like bud. The bud stays attached to the parent cell while it grows and develops. When the bud is fully developed, it breaks away from the parent cell and forms a new organism. Budding in yeast is shown in **Figure** 5.14.

Asexual reproduction can be very rapid. This is an advantage for many organisms. It allows them to crowd out other organisms that reproduce more slowly. Bacteria, for example, may divide several times per hour. Under ideal conditions, 100 bacteria can divide to produce millions of bacterial cells in just a few hours! However, most bacteria do not live under ideal conditions. If they did, the entire surface of the planet would soon be covered with them. Instead, their reproduction is kept in check by limited resources, predators, and their own wastes. This is true of most other organisms as well.

FIGURE 5.13

Starfish reproduce by fragmentation. Starfish, however, are also capable of sexual reproduction.

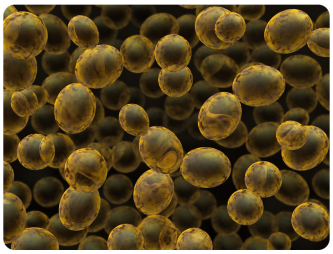

FIGURE 5.14

Yeast reproduces by budding. Both are types of asexual reproduction.

Sexual Reproduction

Sexual reproduction involves two parents. As you can see from **Figure** 5.15, in sexual reproduction, parents produce reproductive cells—called **gametes**—that unite to form an offspring. Gametes are **haploid** cells. This means they contain only half the number of chromosomes found in other cells of the organism. Gametes are produced by a type of cell division called meiosis, which is described in detail below. The process in which two gametes unite is called **fertilization**. The fertilized cell that results is referred to as a **zygote**. A zygote is **diploid** cell, which means that it has twice the number of chromosomes as a gamete.

Mitosis, Meiosis and Sexual Reproduction is discussed at http://www.youtube.com/user/khanacademy#p/c/7A9646BC5110CF64/7/kaSIjIzAtYA.

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Meiosis

The process that produces haploid gametes is meiosis (see **Figure** 5.15). **Meiosis** is a type of cell division in which the number of chromosomes is reduced by half. It occurs only in certain special cells of the organisms. During meiosis, homologous chromosomes separate, and haploid cells form that have only one chromosome from each pair. Two cell divisions occur during meiosis, and a total of four haploid cells are produced. The two cell divisions are called meiosis I and meiosis II. The overall process of meiosis is summarized in **Figure** 5.16. It is also described in detail below. You can watch an animation of meiosis at this link: http://www.youtube.com/watch?v=D1_-mQS_FZ0&feature=related.

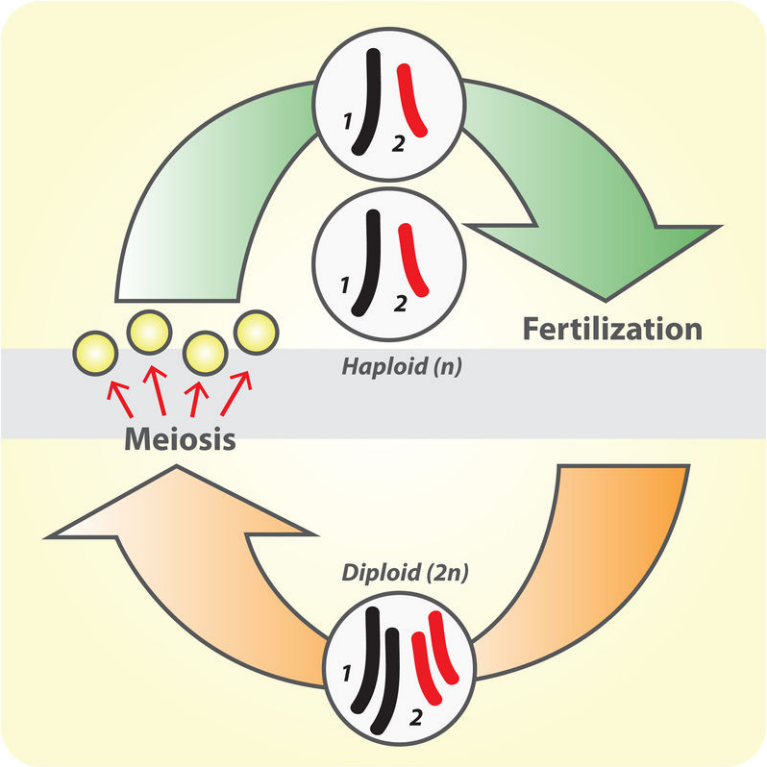


FIGURE 5.15

Cycle of Sexual Reproduction. Sexual reproduction involves the production of haploid gametes by meiosis. This is followed by fertilization and the formation of a diploid zygote. The number of chromosomes in a gamete is represented by the letter n. Why does the zygote have 2n, or twice as many, chromosomes?

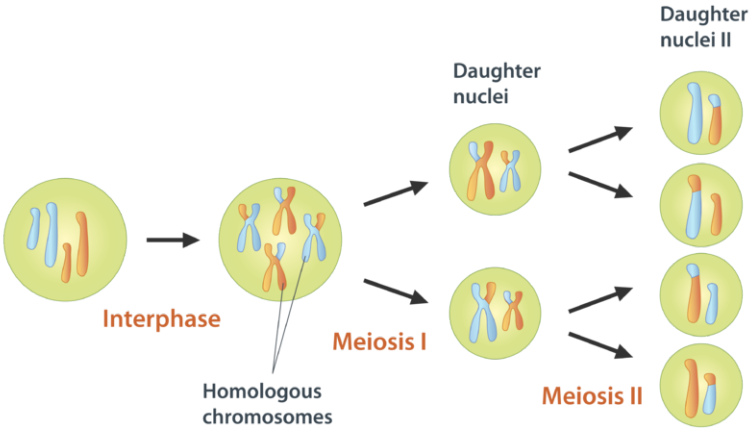


FIGURE 5.16

Overview of Meiosis. During meiosis, homologous chromosomes separate and go to different daughter cells. This diagram shows just the nuclei of the cells.

Phases of Meiosis

Meiosis I begins after DNA replicates during interphase. In both meiosis I and meiosis II, cells go through the same four phases as mitosis. However, there are important differences between meiosis I and mitosis. The flowchart in **Figure** 5.17 shows what happens in both meiosis I and II. You can follow the changes in the flowchart as you read about them below.

The phases of meiosis are discussed at http://www.youtube.com/user/khanacademy#p/c/7A9646BC5110CF64/9/ijLc52LmFQg (27:23).

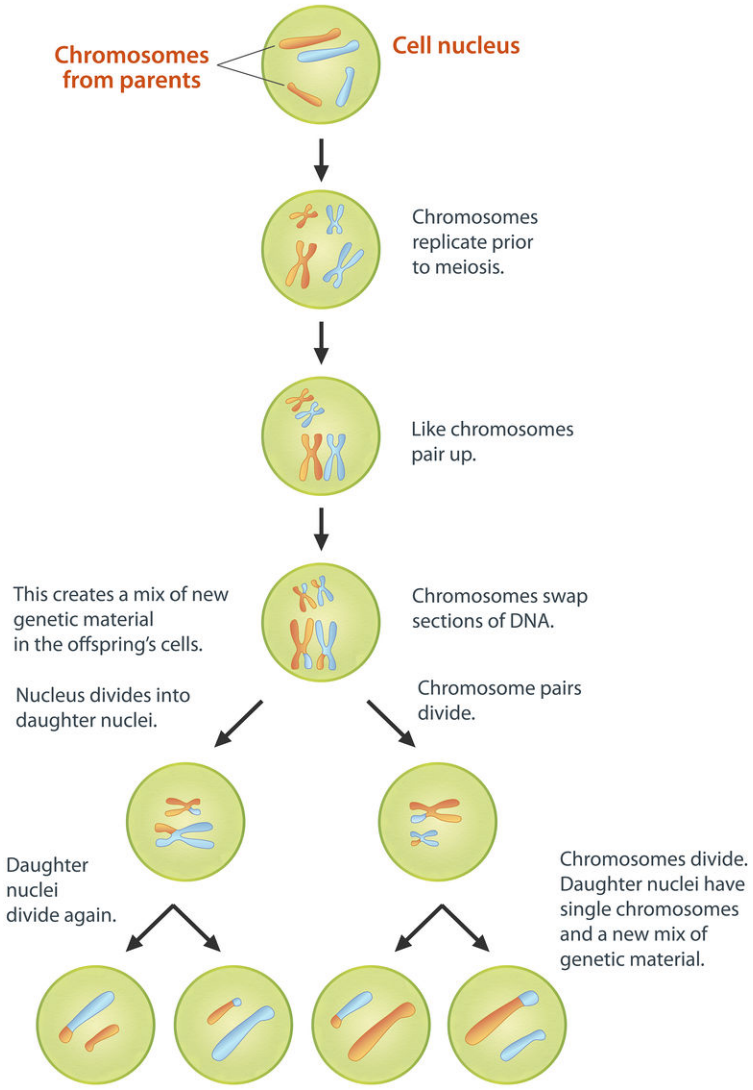
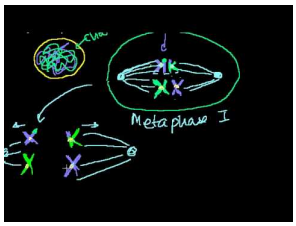


FIGURE 5.17

Phases of Meiosis. This flowchart of meiosis shows meiosis I in greater detail than meiosis II. Meiosis I—but not meiosis II—differs somewhat from mitosis. Compare meiosis I in this flowchart with the earlier figure featuring mitosis. How does meiosis I differ from mitosis?



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Meiosis I

a. Prophase I: The nuclear envelope begins to break down, and the chromosomes condense. Centrioles start moving to opposite poles of the cell, and a spindle begins to form. Importantly, homologous chromosomes pair up, which is unique to prophase I. In prophase of mitosis and meiosis II, homologous chromosomes do not form pairs in this way.
b. Metaphase I: Spindle fibers attach to the paired homologous chromosomes. The paired chromosomes line up

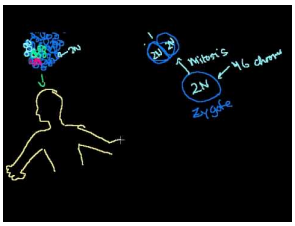
along the equator of the cell. This occurs only in metaphase I. In metaphase of mitosis and meiosis II, it is sister chromatids that line up along the equator of the cell.

c. Anaphase I: Spindle fibers shorten, and the chromosomes of each homologous pair start to separate from each other. One chromosome of each pair moves toward one pole of the cell, and the other chromosome moves toward the opposite pole.
d. Telophase I and Cytokinesis: The spindle breaks down, and new nuclear membranes form. The cytoplasm of the cell divides, and two haploid daughter cells result. The daughter cells each have a random assortment of chromosomes, with one from each homologous pair. Both daughter cells go on to meiosis II.

Meiosis II

a. Prophase II: The nuclear envelope breaks down and the spindle begins to form in each haploid daughter cell from meiosis I. The centrioles also start to separate.
b. Metaphase II: Spindle fibers line up the sister chromatids of each chromosome along the equator of the cell.
c. Anaphase II: Sister chromatids separate and move to opposite poles.
d. Telophase II and Cytokinesis: The spindle breaks down, and new nuclear membranes form. The cytoplasm of each cell divides, and four haploid cells result. Each cell has a unique combination of chromosomes.

Mitosis, Meiosis and Sexual Reproduction is discussed at http://www.youtube.com/user/khanacademy#p/c/7A9646BC5110CF64/7/kaSIjIzAtYA (18:23).



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You can watch an animation of meiosis at this link: http://www.youtube.com/watch?v=D1_-mQS_FZ0&feature=related.

Gametogenesis

At the end of meiosis, four haploid cells have been produced, but the cells are not yet gametes. The cells need to develop before they become mature gametes capable of fertilization. The development of haploid cells into gametes is called **gametogenesis**. Gametogenesis may differ between males and females. Male gametes are called **sperm**. Female gametes are called **eggs**. In human males, for example, the process that produces mature sperm cells is called spermatogenesis. During this process, sperm cells grow a tail and gain the ability to "swim," like the human sperm cell shown in **Figure** 5.18. In human females, the process that produces mature eggs is called oogenesis. Just one egg is produced from the four haploid cells that result from meiosis. The single egg is a very large cell, as you can see from the human egg in **Figure** 5.18.

Sexual Reproduction and Genetic Variation

Sexual reproduction results in offspring that are genetically unique. They differ from both parents and also from each other. This occurs for a number of reasons.

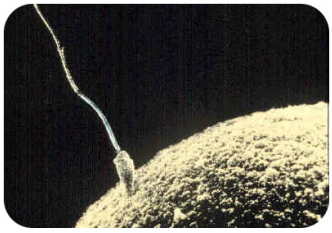

FIGURE 5.18

A human sperm is a tiny cell with a tail. A human egg is much larger. Both cells are mature haploid gametes that are capable of fertilization. What process is shown in this photograph?

- When homologous chromosomes pair up during meiosis I, crossing-over can occur. **Crossing-over** is the exchange of genetic material between homologous chromosomes. It results in new combinations of genes on each chromosome.
- When cells divide during meiosis, homologous chromosomes are randomly distributed to daughter cells, and different chromosomes segregate independently of each other. This called is called **independent assortment**. It results in gametes that have unique combinations of chromosomes.
- In sexual reproduction, two gametes unite to produce an offspring. But which two of the millions of possible gametes will it be? This is likely to be a matter of chance. It is obviously another source of genetic variation in offspring.

All of these mechanisms working together result in an amazing amount of potential variation. Each human couple, for example, has the potential to produce more than 64 trillion genetically unique children. No wonder we are all different!

Sexual Reproduction and Life Cycles

Sexual reproduction occurs in a cycle. Diploid parents produce haploid gametes that unite and develop into diploid adults, which repeat the cycle. This series of life stages and events that a sexually reproducing organism goes through is called its **life cycle**. Sexually reproducing organisms can have different types of life cycles. Three are represented in **Figure** 5.19 and described following sections.

Haploid Life Cycle

The haploid life cycle is the simplest life cycle. It is found in many single-celled organisms. Organisms with a haploid life cycle spend the majority of their lives as haploid gametes. When the haploid gametes fuse, they form a diploid zygote. It quickly undergoes meiosis to produce more haploid gametes that repeat the life cycle.

Diploid Life Cycle

Organisms with a diploid life cycle spend the majority of their lives as diploid adults. When they are ready to reproduce, they undergo meiosis and produce haploid gametes. Gametes then unite in fertilization and form a diploid zygote. The zygote develops into a diploid adult that repeats the life cycle. Can you think of an organism with a diploid life cycle? (*Hint:* What type of life cycle do humans have?)

Alternation of Generations

Organisms that have a life cycle with alternating generations switch back and forth between diploid and haploid stages. Organisms with this type of life cycle include plants, algae, and some protists. These life cycles may be quite

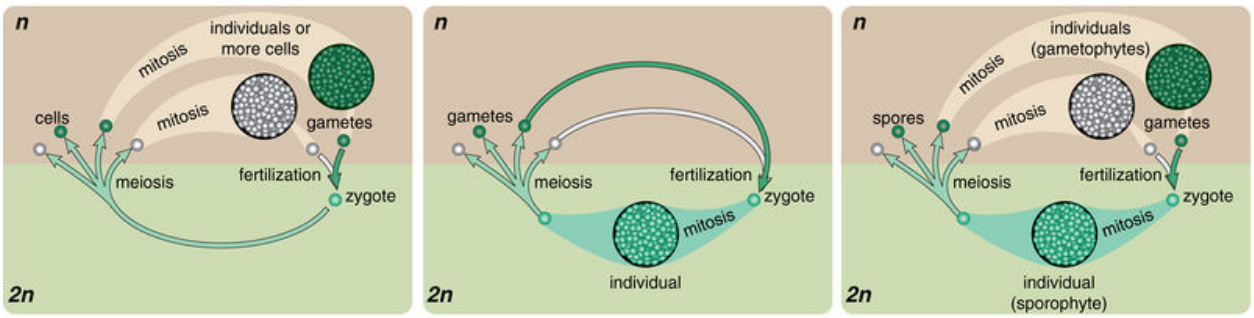

FIGURE 5.19

Life cycles can vary in sexually reproducing organisms. Three types of sexual life cycles are shown here. Do you see how they differ? The letter n indicates haploid stages of the life cycles, and 2n indicates diploid stages.

complicated. You can read about them in later chapters.

Lesson Summary

- Asexual reproduction involves one parent and produces offspring that are genetically identical to each other and to the parent. Sexual reproduction involves two parents and produces offspring that are genetically unique.
- During sexual reproduction, two haploid gametes join in the process of fertilization to produce a diploid zygote. Meiosis is the type of cell division that produces gametes. It involves two cell divisions and produces four haploid cells.
- Sexual reproduction has the potential to produce tremendous genetic variation in offspring. This variation is due to independent assortment and crossing-over during meiosis and random union of gametes during fertilization.
- A life cycle is the sequence of stages an organisms goes through from one generation to the next. Organisms that reproduce sexually can have different types of life cycles, such as haploid or diploid life cycles.

Lesson Review Questions

Recall

1. What are three types of asexual reproduction?
2. Define gamete and zygote. What number of chromosomes does each have?
3. What happens during fertilization?
4. Outline the phases of meiosis.
5. What is a life cycle?

6. What is gametogenesis, and when does it occur?

Apply Concepts

7. Create a diagram to show how crossing-over occurs and how it creates new gene combinations on each chromosome.

8. An adult organism produces gametes that quickly go through fertilization and form diploid zygotes. The zygotes mature into adults, which live for many years. Eventually the adults produce gametes and the cycle repeats. What type of life cycle does this organism have? Explain your answer.

Think Critically

9. Compare and contrast asexual and sexual reproduction.

10. Explain why sexual reproduction results in genetically unique offspring.

11. Explain how meiosis I differs from mitosis.

Points to Consider

In sexually reproducing organisms, parents pass a copy of each type of chromosome to their offspring by producing gametes. When gametes are fertilized and form offspring, each has a unique combination of chromosomes and genes from both parents. The inherited gene combination determines the characteristics of the offspring.

- Is it possible to predict possible gene combinations in offspring from the genes of their parents?
- Can the characteristics of offspring be predicted from the characteristics of their parents?

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5.4 References

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CHAPTER 6 Gregor Mendel and Genetics

Chapter Outline

These purple-flowered plants are not just pretty to look at. Plants like these led to a huge leap forward in biology. The plants are common garden peas, and they were studied in the mid-1800s by an Austrian monk named Gregor Mendel. With his careful experiments, Mendel uncovered the secrets of heredity, or how parents pass characteristics to their offspring. You may not care much about heredity in pea plants, but you probably care about your own heredity. Mendel's discoveries apply to you as well as to peas—and to all other living things that reproduce sexually. In this chapter, you will read about Mendel's experiments and the secrets of heredity that he discovered.

6.1 Mendel's Investigations

Lesson Objectives

- Explain why and how Mendel studied pea plants.
- Describe the results of Mendel's experiments.
- State Mendel's laws of segregation and independent assortment.
- Outline the genetics of inheritance.

Vocabulary

allele one of two or more different versions of the same gene

dominant allele allele that masks the presence of another allele for the same gene when they occur together in a heterozygote

genetics the science of heredity

genotype alleles an individual inherits at a particular genetic locus

heterozygote organism that inherits two different alleles for a given gene

homozygote organism that inherits two alleles of the same type for a given gene

hybrid offspring that results from a cross between two different types of parents

law of independent assortment Mendel's second law stating that factors controlling different characteristics are inherited independently of each other

law of segregation Mendel's first law stating that the two factors controlling a characteristics separate and go to different gametes

locus position of a gene on a chromosome

phenotype characteristics of an organism that depend on how the organism's genotype is expressed

pollen tiny grains that bear the male gametes of seed plants and transfer sperm to female reproductive structures

pollination fertilization in plants in which pollen is transferred to female gametes in an ovary

recessive allele allele that is masked by the presence of another allele for the same gene when they occur together in a heterozygote

Introduction

People have long known that the characteristics of living things are similar in parents and their offspring. Whether it's the flower color in pea plants or nose shape in people, it is obvious that offspring resemble their parents. However, it wasn't until the experiments of Gregor Mendel that scientists understood how characteristics are inherited. Mendel's discoveries formed the basis of **genetics**, the science of heredity. That's why Mendel is often called the "father of genetics." It's not common for a single researcher to have such an important impact on science. The importance of Mendel's work was due to three things: a curious mind, sound scientific methods, and good luck. You'll see why when you read about Mendel's experiments.

An introduction to heredity can be seen at http://www.youtube.com/user/khanacademy#p/c/7A9646BC5110CF64/12/eEUvRrhmcxM (17:27).

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Mendel and His Pea Plants

Gregor Mendel was born in 1822 and grew up on his parents' farm in Austria. He did well in school and became a monk. He also went to the University of Vienna, where he studied science and math. His professors encouraged him to learn science through experimentation and to use math to make sense of his results. Mendel is best known for his experiments with the pea plant *Pisum sativum* (see **Figure** 6.1). You can watch a video about Mendel and his research at the following link: http://www.metacafe.com/watch/hl-19246625/milestones_in_science_engineering_gregor_mendel_and_classical_genetics/.

FIGURE 6.1

Gregor Mendel. The Austrian monk Gregor Mendel experimented with pea plants. He did all of his research in the garden of the monastery where he lived.

Blending Theory of Inheritance

During Mendel's time, the blending theory of inheritance was popular. This is the theory that offspring have a blend, or mix, of the characteristics of their parents. Mendel noticed plants in his own garden that weren't a blend of the parents. For example, a tall plant and a short plant had offspring that were either tall or short but not medium in height. Observations such as these led Mendel to question the blending theory. He wondered if there was a different underlying principle that could explain how characteristics are inherited. He decided to experiment with pea plants to find out. In fact, Mendel experimented with almost 30,000 pea plants over the next several years! At the following link, you can watch an animation in which Mendel explains how he arrived at his decision to study inheritance in pea plants: http://www.dnalc.org/view/16170-Animation-3-Gene-s-don-t-blend-.html.

Why Study Pea Plants?

Why did Mendel choose common, garden-variety pea plants for his experiments? Pea plants are a good choice because they are fast growing and easy to raise. They also have several visible characteristics that may vary. These characteristics, which are shown in **Figure** 6.2, include seed form and color, flower color, pod form and color, placement of pods and flowers on stems, and stem length. Each characteristic has two common values. For example, seed form may be round or wrinkled, and flower color may be white or purple (violet).

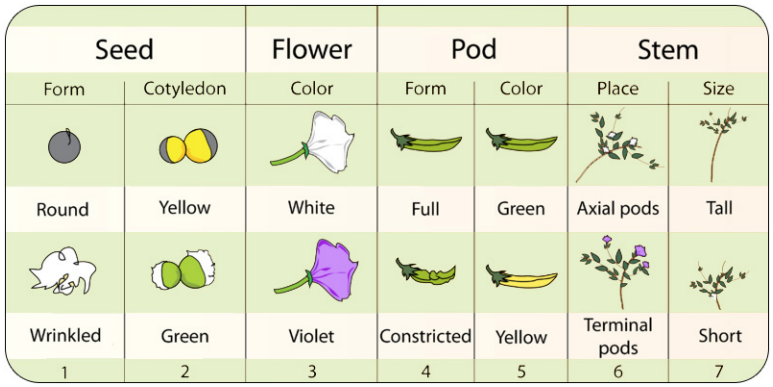

FIGURE 6.2

Mendel investigated seven different characteristics in pea plants. In this chart, cotyledons refer to the tiny leaves inside seeds. Axial pods are located along the stems. Terminal pods are located at the ends of the stems.

Controlling Pollination

To research how characteristics are passed from parents to offspring, Mendel needed to control pollination. **Pollination** is the fertilization step in the sexual reproduction of plants. **Pollen** consists of tiny grains that are the male gametes of plants. They are produced by a male flower part called the anther (see **Figure** 6.3). Pollination occurs when pollen is transferred from the anther to the stigma of the same or another flower. The stigma is a female part of a flower. It passes the pollen grains to female gametes in the ovary.

Pea plants are naturally self-pollinating. In self-pollination, pollen grains from anthers on one plant are transferred to stigmas of flowers on the same plant. Mendel was interested in the offspring of two different parent plants, so he had to prevent self-pollination. He removed the anthers from the flowers of some of the plants in his experiments. Then he pollinated them by hand with pollen from other parent plants of his choice. When pollen from one plant fertilizes another plant of the same species, it is called cross-pollination. The offspring that result from such a cross are called **hybrids.**

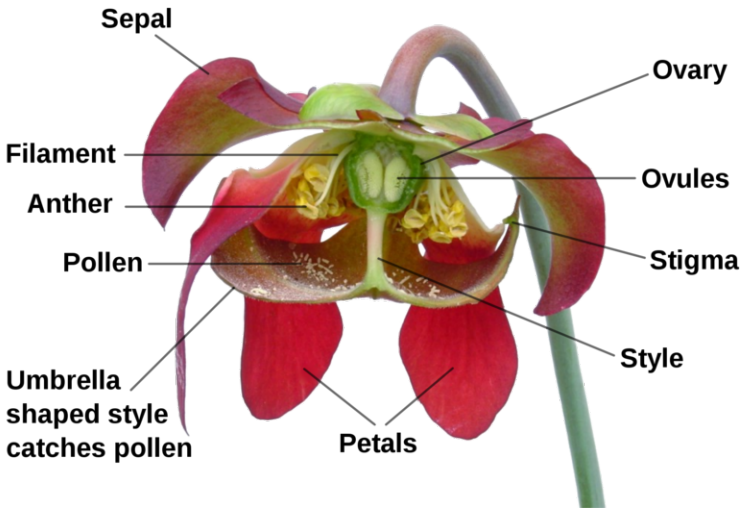

FIGURE 6.3

Flowers are the reproductive organs of plants. Each pea plant flower has both male and female parts. The anther is part of the stamen, the male structure that produces male gametes (pollen). The stigma is part of the pistil, the female structure that produces female gametes and guides the pollen grains to them. The stigma receives the pollen grains and passes them to the ovary, which contains female gametes.

Mendel's First Set of Experiments

At first, Mendel experimented with just one characteristic at a time. He began with flower color. As shown in **Figure** 6.4, Mendel cross-pollinated purple- and white-flowered parent plants. The parent plants in the experiments are referred to as the P (for parent) generation. You can explore an interactive animation of Mendel's first set of experiments at this link: http://www2.edc.org/weblabs/Mendel/mendel.html.

F1 and F2 Generations

The offspring of the P generation are called the F1 (for filial, or "offspring") generation. As you can see from **Figure** 6.4, all of the plants in the F1 generation had purple flowers. None of them had white flowers. Mendel wondered what had happened to the white-flower characteristic. He assumed some type of inherited factor produces white flowers and some other inherited factor produces purple flowers. Did the white-flower factor just disappear in the F1 generation? If so, then the offspring of the F1 generation—called the F2 generation—should all have purple flowers like their parents. To test this prediction, Mendel allowed the F1 generation plants to self-pollinate. He was surprised by the results. Some of the F2 generation plants had white flowers. He studied hundreds of F2 generation plants, and for every three purple-flowered plants, there was an average of one white-flowered plant.

Law of Segregation

Mendel did the same experiment for all seven characteristics. In each case, one value of the characteristic disappeared in the F1 plants and then showed up again in the F2 plants. And in each case, 75 percent of F2 plants had one value of the characteristic and 25 percent had the other value. Based on these observations, Mendel formulated his first law of inheritance. This law is called the **law of segregation**. It states that there are two factors controlling a given characteristic, one of which dominates the other, and these factors separate and go to different gametes when a parent reproduces.

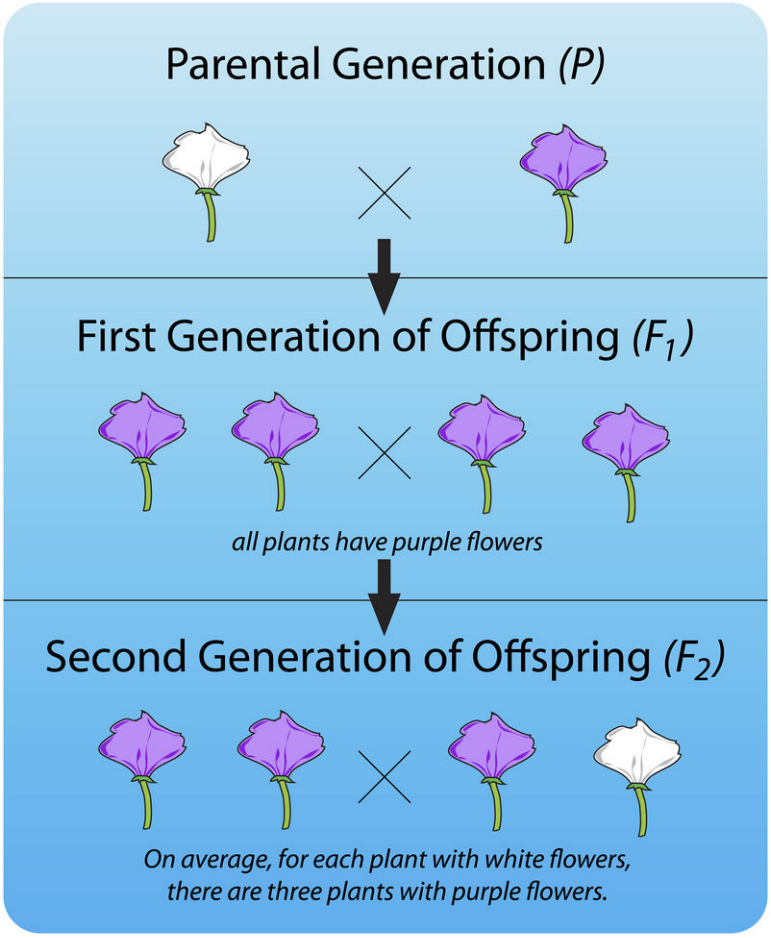

FIGURE 6.4

This diagram shows Mendel's first experiment with pea plants. The F1 generation results from cross-pollination of two parent (P) plants. The F2 generation results from self-pollination of F1 plants.

Mendel's Second Set of Experiments

Mendel wondered whether different characteristics are inherited together. For example, are purple flowers and tall stems always inherited together? Or do these two characteristics show up in different combinations in offspring? To answer these questions, Mendel next investigated two characteristics at a time. For example, he crossed plants with yellow round seeds and plants with green wrinkled seeds. The results of this cross are shown in **Figure** 6.5.

F1 and F2 Generations

In this set of experiments, Mendel observed that plants in the F1 generation were all alike. All of them had yellow and round seeds like one of the two parents. When the F1 generation plants self-pollinated, however, their offspring—the F2 generation—showed all possible combinations of the two characteristics. Some had green round seeds, for example, and some had yellow wrinkled seeds. These combinations of characteristics were not present in the F1 or P generations.

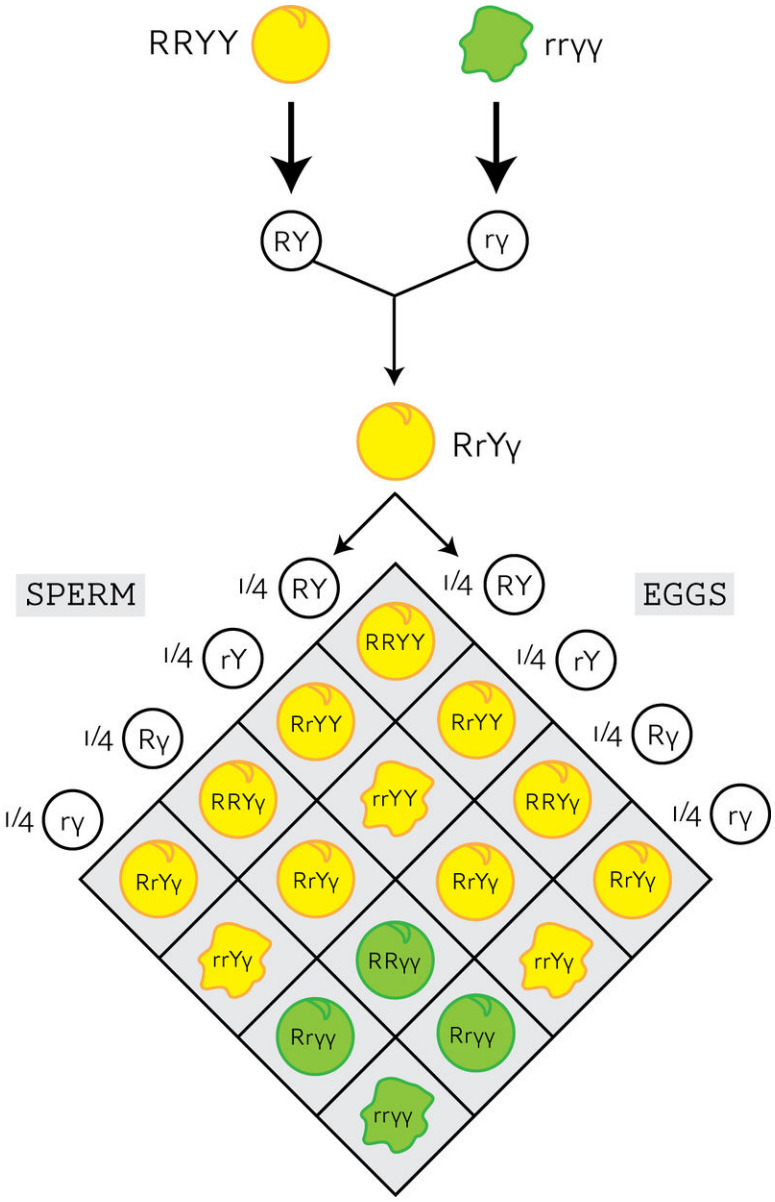

FIGURE 6.5

This chart represents Mendel's second set of experiments. It shows the outcome of a cross between plants that differ in seed color (yellow or green) and seed form (shown here with a smooth round appearance or wrinkled appearance). The letters R, r, Y, and y represent genes for the characteristics Mendel was studying. Mendel didn't know about genes, however. Genes would not be discovered until several decades later. This experiment demonstrates that 9/16 were round yellow, 3/16 were wrinkled yellow, 3/16 were round green, and 1/16 were wrinkled green.

Law of Independent Assortment

Mendel repeated this experiment with other combinations of characteristics, such as flower color and stem length. Each time, the results were the same as those in **Figure** 6.5. The results of Mendel's second set of experiments led to his second law. This is the **law of independent assortment**. It states that factors controlling different characteristics are inherited independently of each other.

Mendel's Laws and Genetics

You might think that Mendel's discoveries would have made a big impact on science as soon as he made them. But you would be wrong. Why? Mendel never published his work. Charles Darwin published his landmark book on evolution in 1869, not long after Mendel had discovered his laws, but Darwin knew nothing of Mendel's discoveries.

As a result, Darwin didn't understand heredity. This made his arguments about evolution less convincing to many people. This example shows why it is important for scientists to communicate the results of their investigations.

Rediscovering Mendel's Work

Mendel's work was virtually unknown until 1900. Then, in that year, three different European scientists—named DeVries, Correns, and Tschermak—independently arrived at Mendel's laws. All three had done experiments similar to Mendel's. They came to the same conclusions that he had drawn almost half a century earlier. Only then was Mendel's actual work rediscovered. As scientists learned more about heredity over the next few decades, they were able to describe Mendel's ideas about inheritance in terms of genes. In this way, the field of genetics was born. At the link that follows, you can watch an animation of Mendel explaining his laws of inheritance in genetic terms. http://www.dnalc.org/view/16182-Animation-4-Some-genes-are-dominant-.html

Genetics of Inheritance

Today, we known that characteristics of organisms are controlled by genes on chromosomes (see **Figure** 6.6). The position of a gene on a chromosome is called its **locus**. In sexually reproducing organisms, each individual has two copies of the same gene. One copy comes from each parent. The gene for a characteristic may have different versions. The different versions are called **alleles**. For example, in pea plants, there is a purple-flower allele (B) and a white-flower allele (b). Different alleles account for much of the variation in the characteristics of organisms.

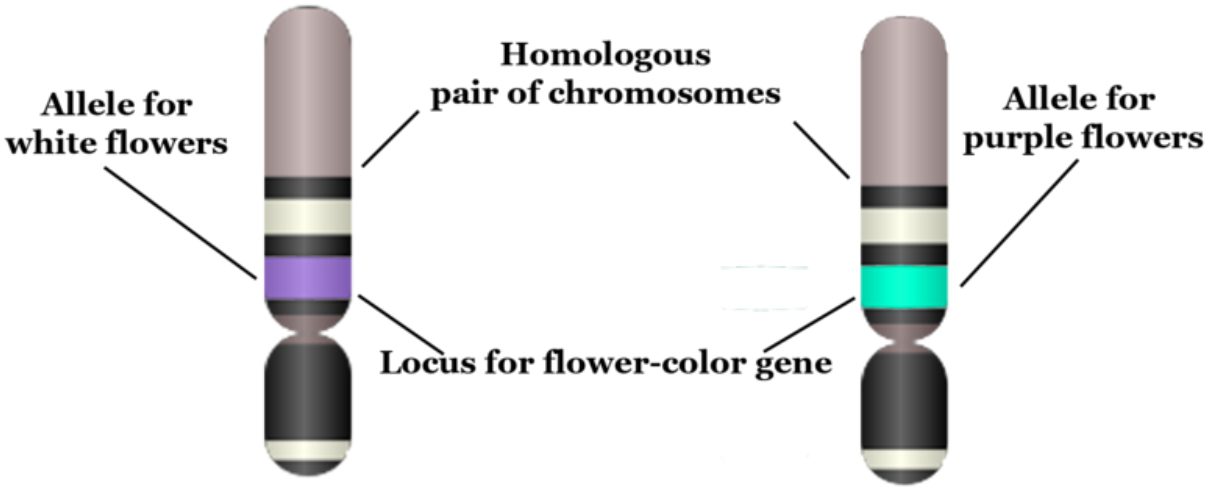

FIGURE 6.6

Chromosome, Gene, Locus, and Allele. This diagram shows how the concepts of chromosome, gene, locus, and allele are related. What is the different between a gene and a locus? Between a gene and an allele?

During meiosis, homologous chromosomes separate and go to different gametes. Thus, the two alleles for each gene also go to different gametes. At the same time, different chromosomes assort independently. As a result, alleles for different genes assort independently as well. In these ways, alleles are shuffled and recombined in each parent's gametes.

Genotype and Phenotype

When gametes unite during fertilization, the resulting zygote inherits two alleles for each gene. One allele comes from each parent. The alleles an individual inherits make up the individual's **genotype**. The two alleles may be the same or different. As shown in **Table** 6.1, an organism with two alleles of the same type (*BB* or *bb*) is called a **homozygote**. An organism with two different alleles (*Bb*) is called a **heterozygote**.

TABLE 6.1: Genetics of Flower Color in Pea Plants

Alleles	Genotypes	Phenotypes
	BB (homozygote)	purple flowers
B (purple)	*Bb* (heterozygote)	purple flowers
b (white)	*bb* (homozygote)	white flowers

Table 6.2 There are two alleles, *B* and *b*, that control flower color in pea plants. This results in three possible genotypes. Why are there only two phenotypes?

The expression of an organism's genotype produces its **phenotype**. The phenotype refers to the organism's characteristics, such as purple or white flowers. As you can see from **Table** 6.1, different genotypes may produce the same phenotype. For example, *BB* and *Bb* genotypes both produce plants with purple flowers. Why does this happen? In a *Bb* heterozygote, only the *B* allele is expressed, so the *b* allele doesn't influence the phenotype. In general, when only one of two alleles is expressed in the phenotype, the expressed allele is called the **dominant allele**. The allele that isn't expressed is called the **recessive allele**.

Lesson Summary

- Gregor Mendel experimented with pea plants to learn how characteristics are passed from parents to offspring.
- First, Mendel researched one characteristic at a time. This led to his law of segregation. This law states that each characteristic is controlled by two factors, which separate and go to different gametes when an organism reproduces.
- Then Mendel researched two characteristics at a time. This led to his law of independent assortment. This law states that the factors controlling different characteristics are inherited independently of each other.
- Mendel's work was rediscovered in 1900. Soon after that, genes and alleles were discovered. This allowed Mendel's laws to be stated in terms of the inheritance of alleles.
- *Gregor Mendel - From the Garden to the Genome* can be viewed at http://www.youtube.com/watch?v=6OPJnO9W_rQ (30.23).

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Lesson Review Questions

Recall

1. What is the blending theory of inheritance? Why did Mendel question this theory?
2. List the seven characteristics that Mendel investigated in pea plants.
3. How did Mendel control pollination in pea plants?
4. Describe in general terms Mendel's first set of experiments.
5. What was Mendel investigating with his second set of experiments? What was the outcome?
6. State Mendel's two laws.

Apply Concepts

7. Assume you are investigating the inheritance of stem length in pea plants. You cross-pollinate a short-stemmed plant with a long-stemmed plant. All of the offspring have long stems. Then, you let the offspring self-pollinate. Describe the stem lengths you would expect to find in the second generation of offspring.
8. If a purple-flowered, short-stemmed plant is crossed with a white-flowered, long-stemmed plant, would all of the purple-flowered offspring also have short stems? Why or why not?

Think Critically

9. If Darwin knew of Mendel's work, how might it have influenced his theory of evolution? Do you think this would have affected how well Darwin's work was accepted?
10. Explain Mendel's laws in genetic terms, that is, in terms of chromosomes, genes, and alleles.
11. Explain the relationship between genotype and phenotype. How can one phenotype result from more than one genotype?

Points to Consider

With his first set of experiments, Mendel found that characteristics appear to skip generations. With his second set of experiments, he found that different characteristics are inherited independently of one another.

- Why would this information be useful? Can you think of a practical application of Mendel's laws?
- Could Mendel's laws be used to predict the characteristics of the offspring of a given set of parents? How do you think this might be done?

6.2 Mendelian Inheritance

Lesson Objectives

- Define probability.
- Explain how probability is related to inheritance.
- Describe how to use a Punnett square.
- Explain how Mendel interpreted the results of his experiments.
- Describe complex patterns of inheritance.

Vocabulary

codominance relationship between two alleles for the same gene in which both alleles are expressed equally in the phenotype of the heterozygote

incomplete dominance relationship between the alleles for a gene in which one allele is only partly dominant to the other allele so an intermediate phenotype results

polygenic characteristic characteristic, or trait, controlled by more than one gene, each of which may have two or more alleles

probability the likelihood, or chance, than a certain event will occur

Punnett square chart for determining the expected percentages of different genotypes in the offspring of two parents

Introduction

Assume you are a plant breeder trying to develop a new variety of plant that is more useful to humans. You plan to cross-pollinate an insect-resistant plant with a plant that grows rapidly. Your goal is to produce a variety of plant that is both insect resistant and fast growing. What percent of the offspring would you expect to have both characteristics? Mendel's laws can be used to find out. However, to understand how Mendel's laws can be used in this way, you first need to know about probability.

Probability

Probability is the likelihood, or chance, that a certain event will occur. The easiest way to understand probability is with coin tosses (see **Figure** 6.7). When you toss a coin, the chance of a head turning up is 50 percent. This is because a coin has only two sides, so there is an equal chance of a head or tail turning up on any given toss.

FIGURE 6.7

Tossing a Coin. Competitions often begin with the toss of a coin. Why is this a fair way to decide who goes first? If you choose heads, what is the chance that the toss will go your way?

If you toss a coin twice, you might expect to get one head and one tail. But each time you toss the coin, the chance of a head is still 50 percent. Therefore, it's quite likely that you will get two or even several heads (or tails) in a row. What if you tossed a coin ten times? You would probably get more or less than the expected five heads. For example, you might get seven heads (70 percent) and three tails (30 percent). The more times you toss the coin, however, the closer you will get to 50 percent heads. For example, if you tossed a coin 1000 times, you might get 510 heads and 490 tails.

Probability and Inheritance

The same rules of probability in coin tossing apply to the main events that determine the genotypes of offspring. These events are the formation of gametes during meiosis and the union of gametes during fertilization.

Probability and Gamete Formation

How is gamete formation like tossing a coin? Consider Mendel's purple-flowered pea plants again. Assume that a plant is heterozygous for the flower-color allele, so it has the genotype *Bb* (see **Figure** 6.8). During meiosis, homologous chromosomes—and the alleles they carry—segregate and go to different gametes. Therefore, when the *Bb* pea plant forms gametes, the *B* and *b* alleles segregate and go to different gametes. As a result, half the gametes produced by the *Bb* parent will have the *B* allele and half will have the *b* allele. Based on the rules of probability, any given gamete of this parent has a 50 percent chance of having the *B* allele and a 50 percent chance of having the *b* allele.

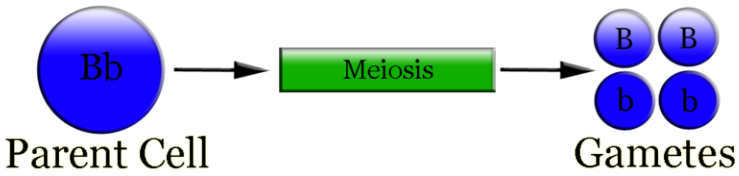


FIGURE 6.8

Formation of Gametes. Paired alleles always separate and go to different gametes during meiosis.

Probability and Fertilization

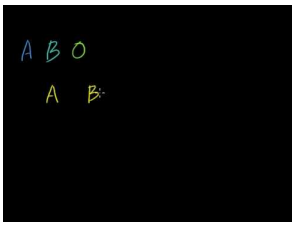
Which of these gametes joins in fertilization with the gamete of another parent plant? This is a matter of chance, like tossing a coin. Thus, we can assume that either type of gamete—one with the *B* allele or one with the *b* allele—has an equal chance of uniting with any of the gametes produced by the other parent. Now assume that the other parent

is also *Bb*. If gametes of two *Bb* parents unite, what is the chance of the offspring having one of each allele like the parents (*Bb*)? What is the chance of them having a different combination of alleles than the parents (either *BB* or *bb*)? To answer these questions, geneticists use a simple tool called a Punnett square.

Using a Punnett Square

A **Punnett square** is a chart that allows you to easily determine the expected percents of different genotypes in the offspring of two parents. An example of a Punnett square for pea plants is shown in **Figure** 6.9. In this example, both parents are heterozygous for flower color (*Bb*). The gametes produced by the male parent are at the top of the chart, and the gametes produced by the female parent are along the side. The different possible combinations of alleles in their offspring are determined by filling in the cells of the Punnett square with the correct letters (alleles). At the link below, you can watch an animation in which Reginald Punnett, inventor of the Punnett square, explains the purpose of his invention and how to use it. http://www.dnalc.org/view/16192-Animation-5-Genetic-inheritance-follows-rules-.html

An explanation of Punnett squares can be viewed at http://www.youtube.com/user/khanacademy#p/c/7A9646BC5110CF64/13/D5ymMYcLtv0 (25:16).



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An example of the use of a Punnett square can be viewed at http://www.youtube.com/watch?v=nsHZbgOmVwg&feature=related (5:40).

Predicting Offspring Genotypes

In the cross shown in **Figure** 6.9, you can see that one out of four offspring (25 percent) has the genotype *BB*, one out of four (25 percent) has the genotype *bb*, and two out of four (50 percent) have the genotype *Bb*. These percents of genotypes are what you would expect in any cross between two heterozygous parents. Of course, when just four offspring are produced, the actual percents of genotypes may vary by chance from the expected percents. However, if you considered hundreds of such crosses and thousands of offspring, you would get very close to the expected results—just like tossing a coin.

Predicting Offspring Phenotypes

You can predict the percents of phenotypes in the offspring of this cross from their genotypes. *B* is dominant to *b*, so offspring with either the *BB* or *Bb* genotype will have the purple-flower phenotype. Only offspring with the *bb* genotype will have the white-flower phenotype. Therefore, in this cross, you would expect three out of four (75 percent) of the offspring to have purple flowers and one out of four (25 percent) to have white flowers. These are the same percents that Mendel got in his first experiment.

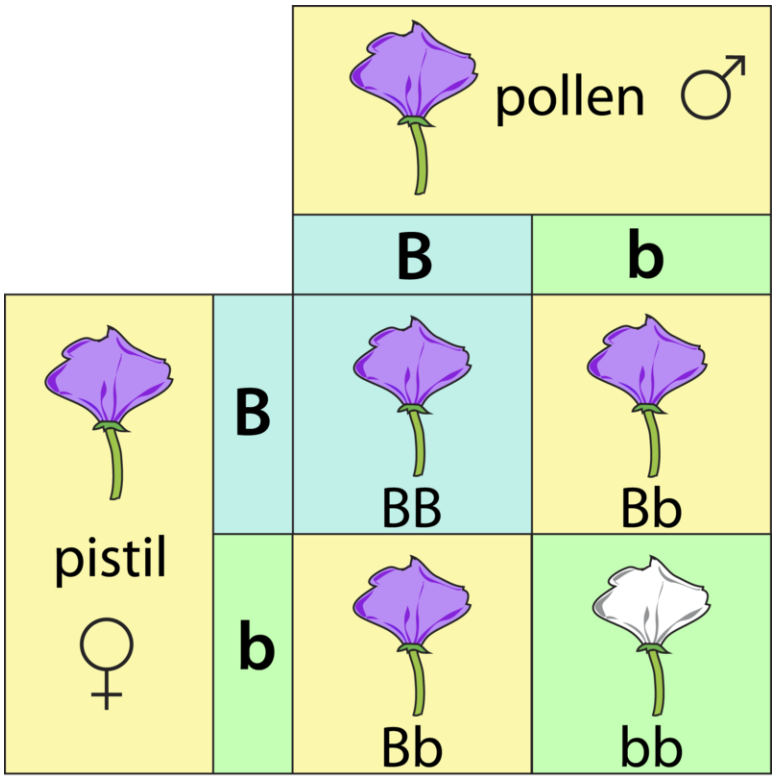

FIGURE 6.9

Punnett Square. This Punnett square shows a cross between two heterozygotes. Do you know where each letter (allele) in all four cells comes from?

Determining Missing Genotypes

A Punnett square can also be used to determine a missing genotype based on the other genotypes involved in a cross. Suppose you have a parent plant with purple flowers and a parent plant with white flowers. Because the *b* allele is recessive, you know that the white-flowered parent must have the genotype *bb*. The purple-flowered parent, on the other hand, could have either the *BB* or the *Bb* genotype. The Punnett square in **Figure** 6.10 shows this cross. The question marks (?) in the chart could be either *B* or *b* alleles.

		White Flowered Parent	
	Parents	**b**	**b**
Purple Flowered Parent	**B**	Bb	Bb
	?	?b	?b

FIGURE 6.10

Punnett Square: Cross Between White-Flowered and Purple-Flowered Pea Plants. This Punnett square shows a cross between a white-flowered pea plant and a purple-flowered pea plant. Can you fill in the missing alleles? What do you need to know about the offspring to complete their genotypes?

Can you tell what the genotype of the purple-flowered parent is from the information in the Punnett square? No; you also need to know the genotypes of the offspring in row 2. What if you found out that two of the four offspring have white flowers? Now you know that the offspring in the second row must have the *bb* genotype. One of their *b* alleles obviously comes from the white-flowered (*bb*) parent, because that's the only allele this parent has. The

other *b* allele must come from the purple-flowered parent. Therefore, the parent with purple flowers must have the genotype *Bb*.

Punnett Square for Two Characteristics

When you consider more than one characteristic at a time, using a Punnett square is more complicated. This is because many more combinations of alleles are possible. For example, with two genes each having two alleles, an individual has four alleles, and these four alleles can occur in 16 different combinations. This is illustrated for pea plants in **Figure** 6.11. In this cross, both parents are heterozygous for pod color (*Gg*) and seed color (*Yy*).

How Mendel Worked Backward to Get Ahead

Mendel used hundreds or even thousands of pea plants in each experiment he did. Therefore, his results were very close to those you would expect based on the rules of probability. For example, in one of his first experiments with flower color, there were 929 plants in the F2 generation. Of these, 705 (76 percent) had purple flowers and 224 (24 percent) had white flowers. Thus, Mendel's results were very close to the 75 percent purple and 25 percent white you would expect by the laws of probability for this type of cross. Of course, Mendel had only phenotypes to work with. He knew nothing about genes and genotypes. Instead, he had to work backward from phenotypes and their percents in offspring to understand inheritance. From the results of his first set of experiments, Mendel realized that there must be two factors controlling each of the characteristics he studied, with one of the factors being dominant to the other. He also realized that the two factors separate and go to different gametes and later recombine in the offspring. This is an example of Mendel's good luck. All of the characteristics he studied happened to be inherited in this way. Mendel also was lucky when he did his second set of experiments. He happened to pick characteristics that are inherited independently of one another. We now know that these characteristics are controlled by genes on nonhomologous chromosomes. What if Mendel had studied characteristics controlled by genes on homologous chromosomes? Would they be inherited together? If so, how do you think this would have affected Mendel's conclusions? Would he have been able to develop his second law of inheritance? To better understand how Mendel interpreted his findings and developed his laws of inheritance, you can visit the following link. It provides an animation in which Mendel explains how he came to understand heredity from his experimental results. http://www.dnalc.org/view/16154-Animation-2-Genes-Come-in-Pairs.html

Non-Mendelian Inheritance

The inheritance of characteristics is not always as simple as it is for the characteristics that Mendel studied in pea plants. Each characteristic Mendel investigated was controlled by one gene that had two possible alleles, one of which was completely dominant to the other. This resulted in just two possible phenotypes for each characteristic. Each characteristic Mendel studied was also controlled by a gene on a different (nonhomologous) chromosome. As a result, each characteristic was inherited independently of the other characteristics. Geneticists now know that inheritance is often more complex than this.

Codominance and Incomplete Dominance

A characteristic may be controlled by one gene with two alleles, but the two alleles may have a different relationship than the simple dominant-recessive relationship that you have read about so far. For example, the two alleles may have a codominant or incompletely dominant relationship. The former is illustrated by the flower in **Figure** 6.12, and the latter in **Figure** 6.13.

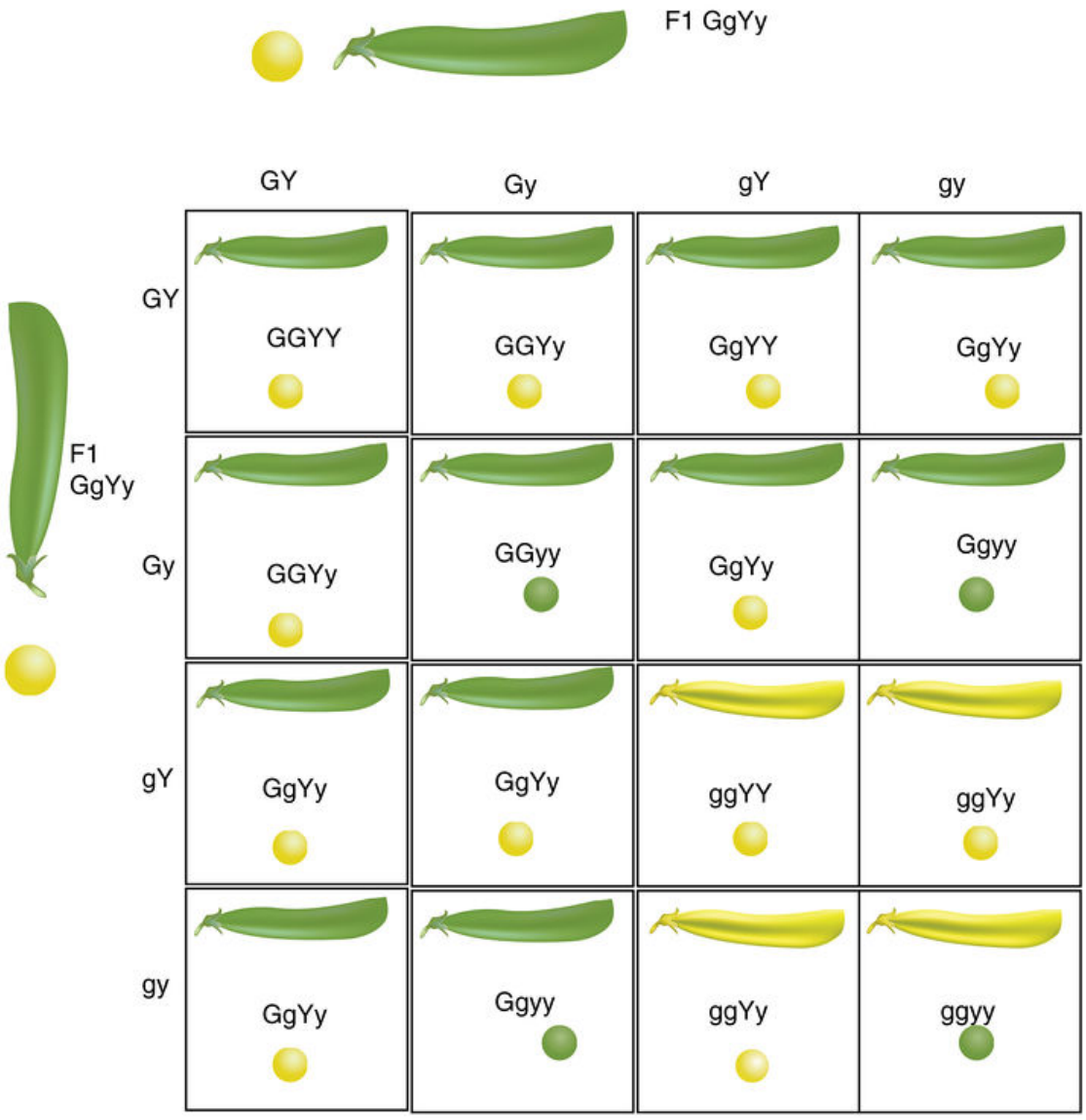

FIGURE 6.11

Punnett Square for Two Characteristics. This Punnett square represents a cross between two pea plants that are heterozygous for two characteristics. G represents the dominant allele for green pod color, and g represents the recessive allele for yellow pod color. Y represents the dominant allele for yellow seed color, and y represents the recessive allele for green seed color.

Codominance

Codominance occurs when both alleles are expressed equally in the phenotype of the heterozygote. The red and white flower in the figure has codominant alleles for red petals and white petals.

Incomplete Dominance

Incomplete dominance occurs when the dominant allele is not completely dominant. Expression of the dominant allele is influenced by the recessive allele, and an intermediate phenotype results. The pink flower in the figure has an incompletely dominant allele for red petals and a recessive allele for white petals.

FIGURE 6.12

Codominance. The flower has red and white petals because of codominance of red-petal and white-petal alleles.

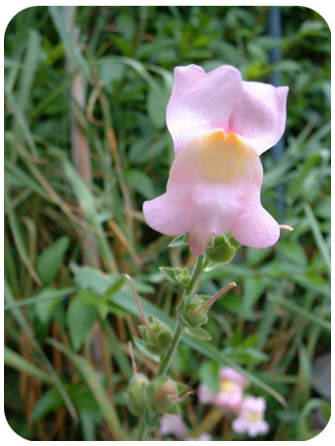

FIGURE 6.13

Incomplete Dominance. The flower has pink petals because of incomplete dominance of a red-petal allele and a recessive white-petal allele.

Multiple Alleles

Many genes have multiple (more than two) alleles. An example is ABO blood type in humans. There are three common alleles for the gene that controls this characteristic. The allele for type A is codominant with the allele for type B, and both alleles are dominant to the allele for type O. Therefore, the possible phenotypes are type A, B, AB, and O. Do you know what genotypes produce these phenotypes?

Polygenic Characteristics

Polygenic characteristics are controlled by more than one gene, and each gene may have two or more alleles. The genes may be on the same chromosome or on nonhomologous chromosomes.

- If the genes are located close together on the same chromosome, they are likely to be inherited together. However, it is possible that they will be separated by crossing-over during meiosis, in which case they may be inherited independently of one another.

- If the genes are on nonhomologous chromosomes, they may be recombined in various ways because of independent assortment.

For these reasons, the inheritance of polygenic characteristics is very complicated. Such characteristics may have many possible phenotypes. Skin color and adult height are examples of polygenic characteristics in humans. Do you have any idea how many phenotypes each characteristic has?

Effects of Environment on Phenotype

Genes play an important role in determining an organism's characteristics. However, for many characteristics, the individual's phenotype is influenced by other factors as well. Environmental factors, such as sunlight and food availability, can affect how genes are expressed in the phenotype of individuals. Here are just two examples:

- Genes play an important part in determining our adult height. However, factors such as poor nutrition can prevent us from achieving our full genetic potential.
- Genes are a major determinant of human skin color. However, exposure to ultraviolet radiation can increase the amount of pigment in the skin and make it appear darker.

Lesson Summary

- Probability is the chance that a certain event will occur. For example, the probability of a head turning up on any given coin toss is 50 percent.
- Probability can be used to predict the chance of gametes and offspring having certain alleles.
- A Punnett square is a chart for determining the expected percents of different genotypes and phenotypes in the offspring of two parents.
- Mendel used the percents of phenotypes in offspring to understand how characteristics are inherited.
- Many characteristics have more complex inheritance patterns than those studied by Mendel. They are complicated by factors such as codominance, incomplete dominance, multiple alleles, and environmental influences.

Lesson Review Questions

Recall

1. Define probability. Apply the term to a coin toss.

2. How is gamete formation like tossing a coin?

3. What is a Punnett square? How is it used?

4. What information must you know to determine the phenotypes of different genotypes for a gene with two alleles?

5. Based on the results of his experiments, what did Mendel conclude about the factors that control characteristics such as flower color?

Apply Concepts

6. Draw a Punnett square of an *Ss* x *ss* cross. The S allele codes for long stems in pea plants and the s allele codes for short stems. If S is dominant to s, what percent of offspring would you expect to have each phenotype?

7. What letter should replace the question marks (?) in this Punnett square? Explain how you know.

	A	A
?	A?	Aa
?	Aa	A?

Think Critically

8. Explain how Mendel used math and probability to understand the results of his experiments.

9. Compare and contrast codominance and incomplete dominance.

10. Mendel investigated stem length, or height, in pea plants. What if he had investigated human height instead? Why would his results have been harder to interpret?

Points to Consider

Like most of the characteristics of living things, the characteristics Mendel studied in pea plants are controlled by genes. All the cells of an organism contain the same genes, because all organisms begin as a single cell. Most of the genes code for proteins.

- How is the information encoded in a gene translated into a protein? Where does this occur, and what processes are involved?
- If cells have the same genes, how do you think different cells arise in an organism? For example, how did you come to have different skin, bone, and blood cells if all of your cells contain the same genes?

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6.3 References

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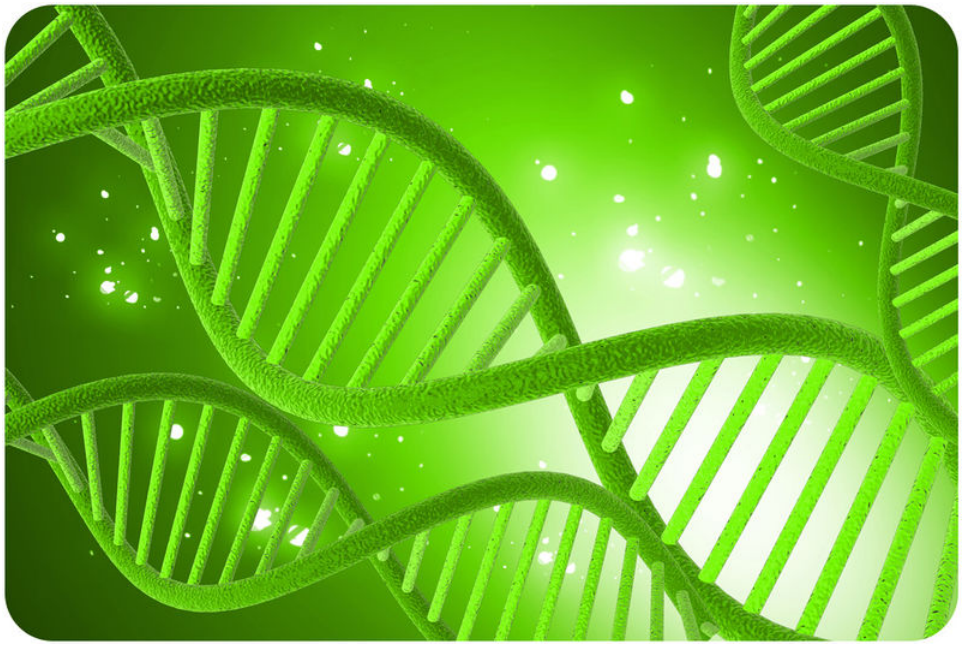
CHAPTER 7 Molecular Genetics: From DNA to Proteins

Chapter Outline



The spiral structure in the picture is a large organic molecule. Can you guess what it is? Here's a hint: molecules like this one determine who you are. They contain genetic information that controls your characteristics. They determine your eye color, facial features, and other physical attributes. What molecule is it?

You probably answered "DNA." Today, it is commonly known that DNA is the genetic material. For a long time, scientists knew such molecules existed. They were aware that genetic information was contained within organic molecules. However, they didn't know which type of molecules play this role. In fact, for many decades, scientists thought that proteins were the molecules that carry genetic information. In this chapter, you will learn how scientists discovered that DNA carries the code of life.

7.1 DNA and RNA

Lesson Objectives

- State the central dogma of molecular biology.
- Outline discoveries that led to knowledge of DNA's structure and function.
- Describe the structure of RNA, and identify the three main types of RNA.

Vocabulary

central dogma of molecular biology doctrine that genetic instructions in DNA are copied by RNA, which carries them to a ribosome where they are used to synthesize a protein (DNA → RNA → protein)

Chargaff's rules observations by Erwin Chargaff that concentrations of the four nucleotide bases differ among species; and that, within a species, the concentrations of adenine and thymine are always about the same and the concentrations of cytosine and guanine are always about the same

messenger RNA (mRNA) type of RNA that copies genetic instructions from DNA in the nucleus and carries them to the cytoplasm

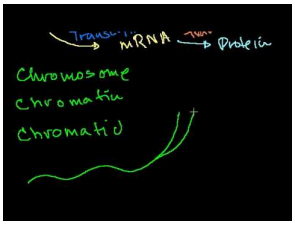
ribosomal RNA (rRNA) type of RNA that helps form ribosomes and assemble proteins

transfer RNA (tRNA) type of RNA that brings amino acids to ribosomes where they are joined together to form proteins

Introduction

Your DNA, or deoxyribonucleic acid, contains the genes that determine who you are. How can this organic molecule control your characteristics? DNA contains instructions for all the proteins your body makes. Proteins, in turn, determine the structure and function of all your cells. What determines a protein's structure? It begins with the sequence of amino acids that make up the protein. Instructions for making proteins with the correct sequence of amino acids are encoded in DNA.

The vocabulary of DNA: chromosomes, chromatids, chromatin, transcription, translation, and replication is discussed at http://www.youtube.com/user/khanacademy#p/c/7A9646BC5110CF64/6/s9HPNwXd9fk (18:23).



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Central Dogma of Molecular Biology

DNA is found in chromosomes. In eukaryotic cells, chromosomes always remain in the nucleus, but proteins are made at ribosomes in the cytoplasm. How do the instructions in DNA get to the site of protein synthesis outside the nucleus? Another type of nucleic acid is responsible. This nucleic acid is RNA, or ribonucleic acid. RNA is a small molecule that can squeeze through pores in the nuclear membrane. It carries the information from DNA in the nucleus to a ribosome in the cytoplasm and then helps assemble the protein. In short:

DNA → RNA → Protein

Discovering this sequence of events was a major milestone in molecular biology. It is called the **central dogma of molecular biology**. You can watch a video about the central dogma and other concepts in this lesson at this link: http://www.youtube.com/watch?v=ZjRCmU0_dhY&feature=fvw (8:07). An overview of protein synthesis can be viewed at http://www.youtube.com/watch?v=-ygpqVr7_xs&feature=related (10:46).

DNA

DNA is the genetic material in your cells. It was passed on to you from your parents and determines your characteristics. The discovery that DNA is the genetic material was another important milestone in molecular biology.

Griffith Searches for the Genetic Material

Many scientists contributed to the identification of DNA as the genetic material. In the 1920s, Frederick Griffith made an important discovery. He was studying two different strains of a bacterium, called R (rough) strain and S (smooth) strain. He injected the two strains into mice. The S strain killed (virulent) the mice, but the R strain did not (nonvirulent) (see **Figure** 7.1). Griffith also injected mice with S-strain bacteria that had been killed by heat. As expected, the killed bacteria did not harm the mice. However, when the dead S-strain bacteria were mixed with live R-strain bacteria and injected, the mice died.

Based on his observations, Griffith deduced that something in the killed S-strain was transferred to the previously harmless R-strain, making the R-strain deadly. What was that something? What type of substance could change the characteristics of the organism that received it?

Avery's Team Makes a Major Contribution

In the early 1940s, a team of scientists led by Oswald Avery tried to answer the question raised by Griffith's results. They inactivated various substances in the S-strain bacteria. They then killed the S-strain bacteria and mixed the remains with live R-strain bacteria. (Keep in mind, the R-strain bacteria usually did not harm the mice.) When they inactivated proteins, the R-strain was deadly to the injected mice. This ruled out proteins as the genetic material. Why? Even without the S-strain proteins, the R-strain was changed, or transformed, into the deadly strain. However, when the researchers inactivated DNA in the S-strain, the R-strain remained harmless. This led to the conclusion that DNA is the substance that controls the characteristics of organisms. In other words, DNA is the genetic material. You can watch an animation about the research of both Griffith and Avery at this link: http://www.dnalc.org/view/16375-Animation-17-A-gene-is-made-of-DNA-.html.

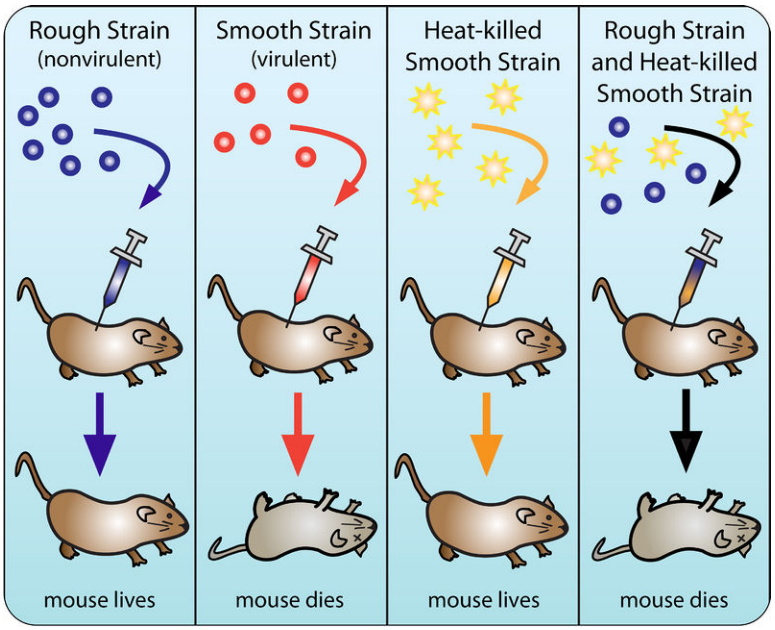


FIGURE 7.1

Griffith's Experimental Results. Griffith showed that a substance could be transferred to harmless bacteria and make them deadly.

Hershey and Chase Seal the Deal

The conclusion that DNA is the genetic material was not widely accepted at first. It had to be confirmed by other research. In the 1950s, Alfred Hershey and Martha Chase did experiments with viruses and bacteria. Viruses are not cells. They are basically DNA inside a protein coat. To reproduce, a virus must insert its own genetic material into a cell (such as a bacterium). Then it uses the cell's machinery to make more viruses. The researchers used different radioactive elements to label the DNA and proteins in viruses. This allowed them to identify which molecule the viruses inserted into bacteria. DNA was the molecule they identified. This confirmed that DNA is the genetic material.

Chargaff Writes the Rules

Other important discoveries about DNA were made in the mid-1900s by Erwin Chargaff. He studied DNA from many different species. He was especially interested in the four different nitrogen bases of DNA: adenine (A), guanine (G), cytosine (C), and thymine (T) (see **Figure** 7.2). Chargaff found that concentrations of the four bases differed from one species to another. However, within each species, the concentration of adenine was always about the same as the concentration of thymine. The same was true of the concentrations of guanine and cytosine. These observations came to be known as **Chargaff's rules**. The significance of the rules would not be revealed until the structure of DNA was discovered.

The Double Helix

After DNA was found to be the genetic material, scientists wanted to learn more about it. James Watson and Francis Crick are usually given credit for discovering that DNA has a double helix shape, like a spiral staircase (see **Figure** 7.3). The discovery was based on the prior work of Rosalind Franklin and other scientists, who had used X rays to learn more about DNA's structure. Franklin and these other scientists have not always been given credit for their contributions. You can learn more about Franklin's work by watching the video at this link: http://www.youtube.com/watch?v=s3whouvZYG8 (7:47).

The double helix shape of DNA, together with Chargaff's rules, led to a better understanding of DNA. DNA, as a

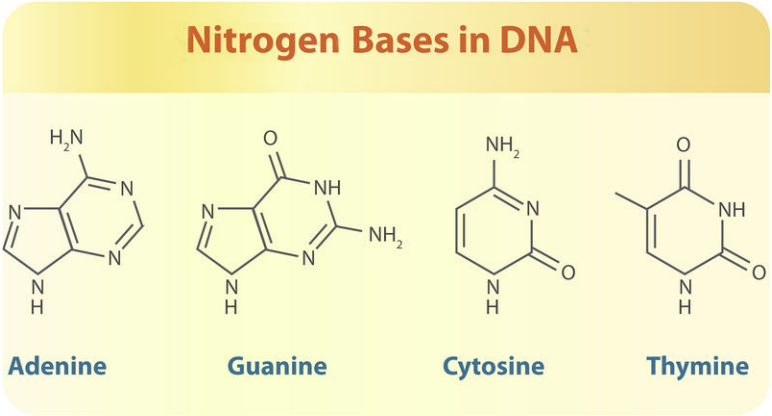


FIGURE 7.2

Nitrogen Bases in DNA. The DNA of all species has the same four nitrogen bases.

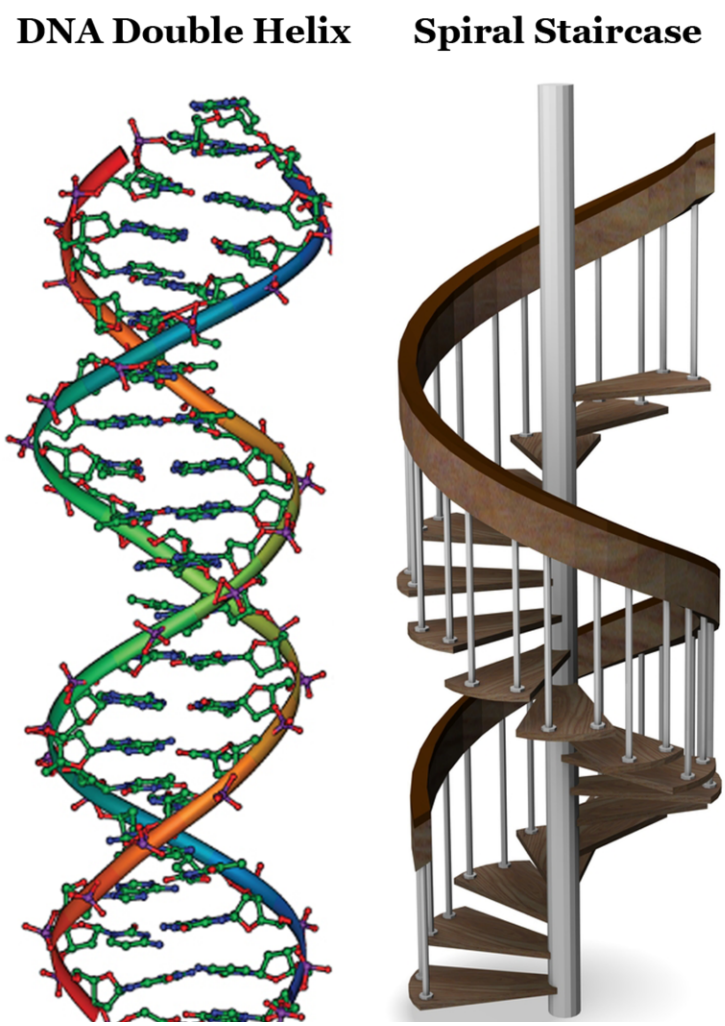


FIGURE 7.3

The DNA molecule has a double helix shape. This is the same basic shape as a spiral staircase. Do you see the resemblance? Which parts of the DNA molecule are like the steps of the spiral staircase?

nucleic acid, is made from nucleotide monomers, and the DNA double helix consists of two polynucleotide chains. Each nucleotide consists of a sugar (deoxyribose), a phosphate group, and a nitrogen-containing base (A, C, G, or T). The sugar-phosphate backbone of the double helix was discussed in the *Chemistry of Life* chapter.

Scientists concluded that bonds (hydrogen bonds) between complementary bases hold together the two polynucleotide chains of DNA. Adenine always bonds with its complementary base, thymine. Cytosine always bonds with its complementary base, guanine. If you look at the nitrogen bases in **Figure** 7.2, you will see why. Adenine and guanine have a two-ring structure. Cytosine and thymine have just one ring. If adenine were to bind with guanine and cytosine with thymine, the distance between the two DNA chains would be variable. However, when a one-ring molecule binds with a two-ring molecule, the distance between the two chains is kept constant. This maintains the uniform shape of the DNA double helix. These *base pairs* (A-T or G-C) stick into the middle of the double helix, forming, in essence, the steps of the spiral staircase.

DNA Replication

Knowledge of DNA's structure helped scientists understand how DNA replicates. DNA replication is the process in which DNA is copied. It occurs during the synthesis (S) phase of the eukaryotic cell cycle. DNA replication begins when an enzyme breaks the bonds between complementary bases in DNA (see **Figure** 7.4). This exposes the bases inside the molecule so they can be "read" by another enzyme and used to build two new DNA strands with complementary bases. The two daughter molecules that result each contain one strand from the parent molecule and one new strand that is complementary to it. As a result, the two daughter molecules are both identical to the parent molecule. The process of DNA replication is actually much more complex than this simple summary. You can see a detailed animation of the process at this link: http://www.youtube.com/watch?v=-mtLXpgjHL0&NR=1 (2:05).

RNA

DNA alone cannot "tell" your cells how to make proteins. It needs the help of RNA, the other main player in the central dogma of molecular biology. Remember, DNA "lives" in the nucleus, but proteins are made on the ribosomes in the cytoplasm. How does the genetic information get from the nucleus to the cytoplasm? RNA is the answer.

RNA vs. DNA

RNA, like DNA, is a nucleic acid. However, RNA differs from DNA in several ways. In addition to being smaller than DNA, RNA also

- consists of one nucleotide chain instead of two,
- contains the nitrogen base uracil (U) instead of thymine,
- contains the sugar ribose instead of deoxyribose.

Types of RNA

There are three main types of RNA, all of which are involved in making proteins.

a. **Messenger RNA (mRNA)** copies the genetic instructions from DNA in the nucleus, and carries them to the cytoplasm.
b. **Ribosomal RNA (rRNA)** helps form ribosomes, where proteins are assembled.
c. **Transfer RNA (tRNA)** brings amino acids to ribosomes, where they are joined together to form proteins.

In the next lesson, you can read in detail how these three types of RNA help cells make proteins.

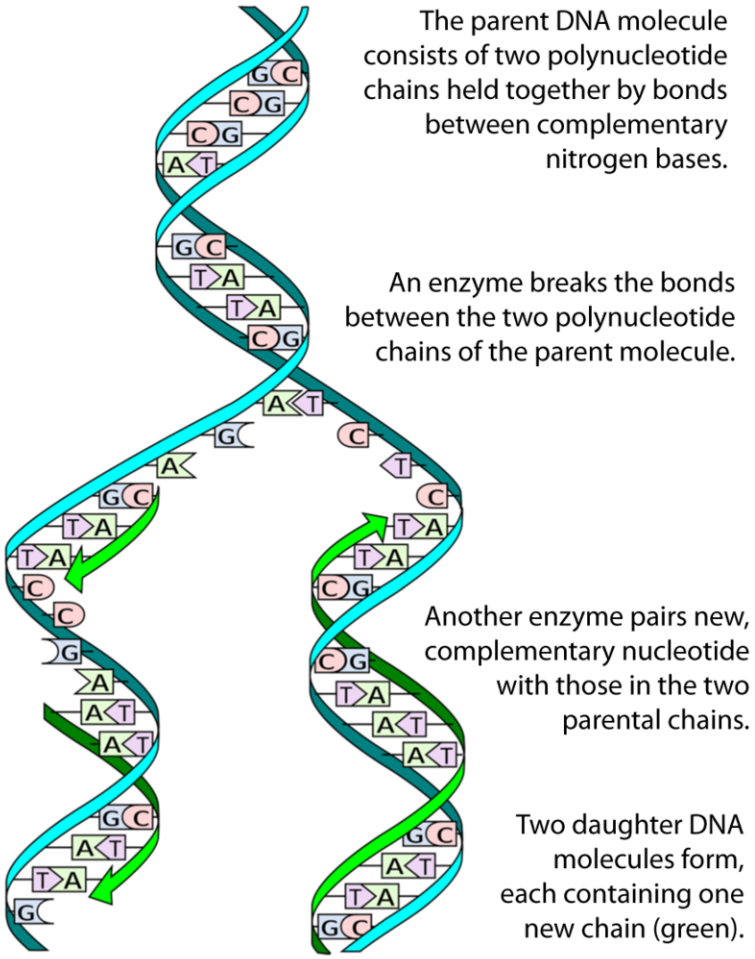


FIGURE 7.4

DNA Replication. DNA replication is a semi-conservative process. Half of the parent DNA molecule is conserved in each of the two daughter DNA molecules.

Lesson Summary

- The central dogma of molecular biology states that DNA contains instructions for making a protein, which are copied by RNA. RNA then uses the instructions to make a protein. In short: *DNA* $\rightarrow$ *RNA* $\rightarrow$ *Protein*.
- The work of several researchers led to the discovery that DNA is the genetic material. Other researchers discovered that DNA has a double helix shape, consisting of two polynucleotide chains held together by bonds between complementary bases.
- RNA differs from DNA in several ways. There three main types of RNA: messenger RNA (mRNA), ribosomal RNA (rRNA), and transfer RNA (tRNA). Each type plays a different in role in making proteins.

Lesson Review Questions

Recall

1. State the central dogma of molecular biology.
2. Outline research that determined that DNA is the genetic material.
3. What are Chargaff's rules?
4. Identify the structure of the DNA molecule.
5. Why is DNA replication said to be semi-conservative?

Apply Concepts

6. Create a diagram that shows how DNA replication occurs.

Think Critically

7. Explain why complementary base pairing is necessary to maintain the double helix shape of the DNA molecule.
8. Compare and contrast DNA and RNA.

Points to Consider

All three types of RNA are needed by cells to make proteins.

- Can you develop a model in which the three types of RNA interact to make a protein?
- How do you think mRNA copies the genetic instructions in DNA? How are these instructions encoded in the DNA molecule?

7.2 Protein Synthesis

Lesson Objectives

- Give an overview of transcription.
- Describe the genetic code.
- Explain how translation occurs.

Vocabulary

codon group of three nitrogen bases in nucleic acids that makes up a code "word" of the genetic code and stands for an amino acid, start, or stop

genetic code universal code of three-base codons that encodes the genetic instructions for the amino acid sequence of proteins

promoter region of a gene where a RNA polymerase binds to initiate transcription of the gene

protein synthesis process in which cells make proteins that includes transcription of DNA and translation of mRNA

transcription process in which genetic instructions in DNA are copied to form a complementary strand of mRNA

translation process in which genetic instructions in mRNA are "read" to synthesize a protein

Introduction

The process in which cells make proteins is called **protein synthesis**. It actually consists of two processes: transcription and translation. Transcription takes place in the nucleus. It uses DNA as a template to make an RNA molecule. RNA then leaves the nucleus and goes to a ribosome in the cytoplasm, where translation occurs. Translation reads the genetic code in mRNA and makes a protein.

Transcription

Transcription is the first part of the central dogma of molecular biology: **DNA** $\rightarrow$ **RNA**. It is the transfer of genetic instructions in DNA to mRNA. During transcription, a strand of mRNA is made that is complementary to a strand of DNA. **Figure** 7.5 shows how this occurs. You can watch an animation of the process at this link: http://www.biostudio.com/d_%20Transcription.htm.

- A detailed video about transcription is available at this link: http://vcell.ndsu.edu/animations/transcription/movie-flash.htm.

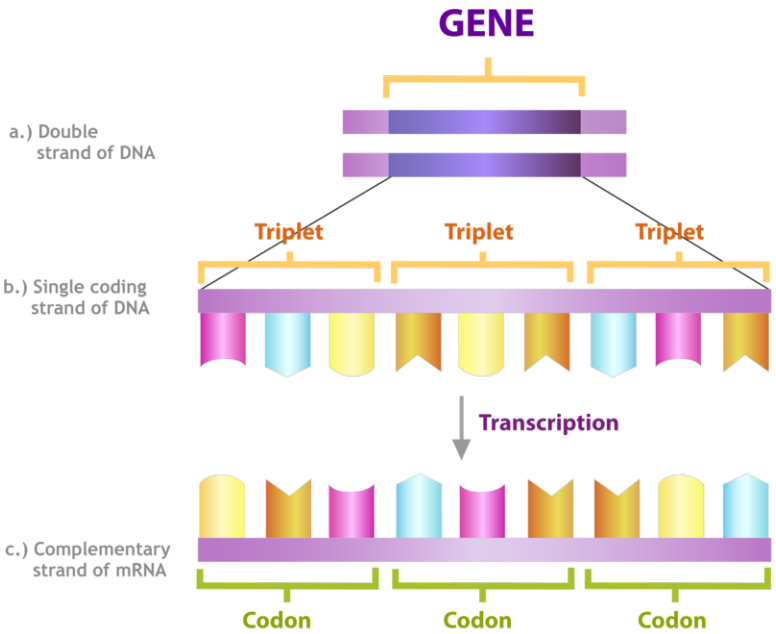


FIGURE 7.5

Overview of Transcription. Transcription uses the sequence of bases in a strand of DNA to make a complementary strand of mRNA. Triplets are groups of three successive nucleotide bases in DNA. Codons are complementary groups of bases in mRNA.

Steps of Transcription

Transcription takes place in three steps: initiation, elongation, and termination. The steps are illustrated in **Figure** 7.6.

a. Initiation is the beginning of transcription. It occurs when the enzyme RNA polymerase binds to a region of a gene called the **promoter**. This signals the DNA to unwind so the enzyme can "read" the bases in one of the DNA strands. The enzyme is ready to make a strand of mRNA with a complementary sequence of bases.
b. Elongation is the addition of nucleotides to the mRNA strand.
c. Termination is the ending of transcription. The mRNA strand is complete, and it detaches from DNA.

Processing mRNA

In eukaryotes, the new mRNA is not yet ready for translation. It must go through more processing before it leaves the nucleus. This may include splicing, editing, and polyadenylation. These processes modify the mRNA in various ways. Such modifications allow a single gene to be used to make more than one protein.

- Splicing removes introns from mRNA (see **Figure** below). Introns are regions that do not code for proteins. The remaining mRNA consists only of regions that do code for proteins, which are called exons. You can watch a video showing splicing in more detail at this link: http://vcell.ndsu.edu/animations/mrnasplicing/movie-flash.htm.
- Editing changes some of the nucleotides in mRNA. For example, the human protein called APOB, which helps transport lipids in the blood, has two different forms because of editing. One form is smaller than the other because editing adds a premature stop signal in mRNA.

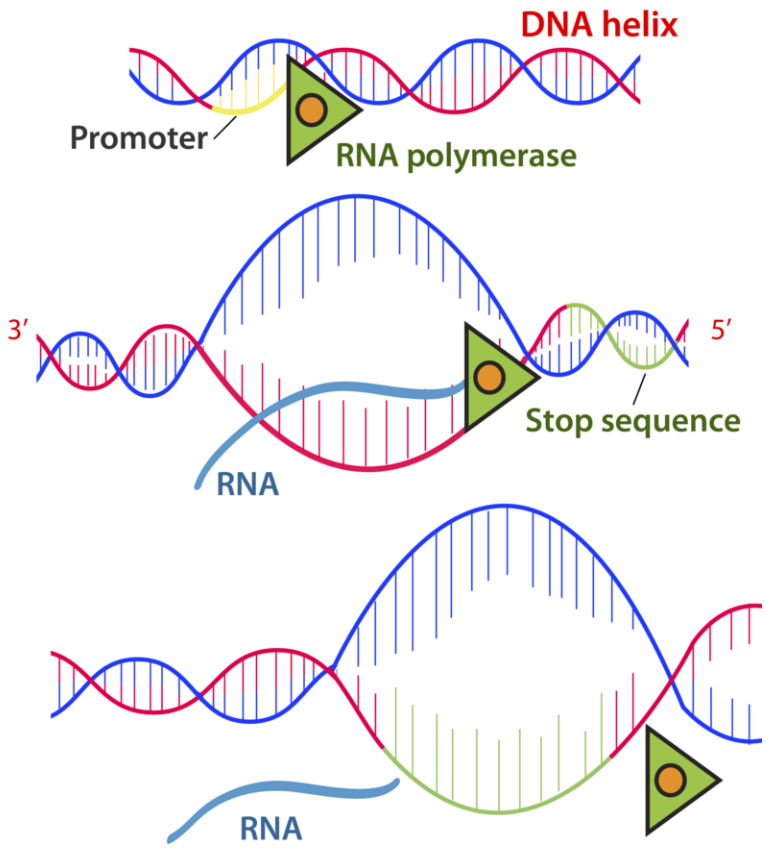


FIGURE 7.6

Steps of Transcription. Transcription occurs in the three steps - initiation, elongation, and termination - shown here.

- Polyadenylation adds a "tail" to the mRNA. The tail consists of a string of As (adenine bases). It signals the end of mRNA. It is also involved in exporting mRNA from the nucleus. In addition, the tail protects mRNA from enzymes that might break it down.

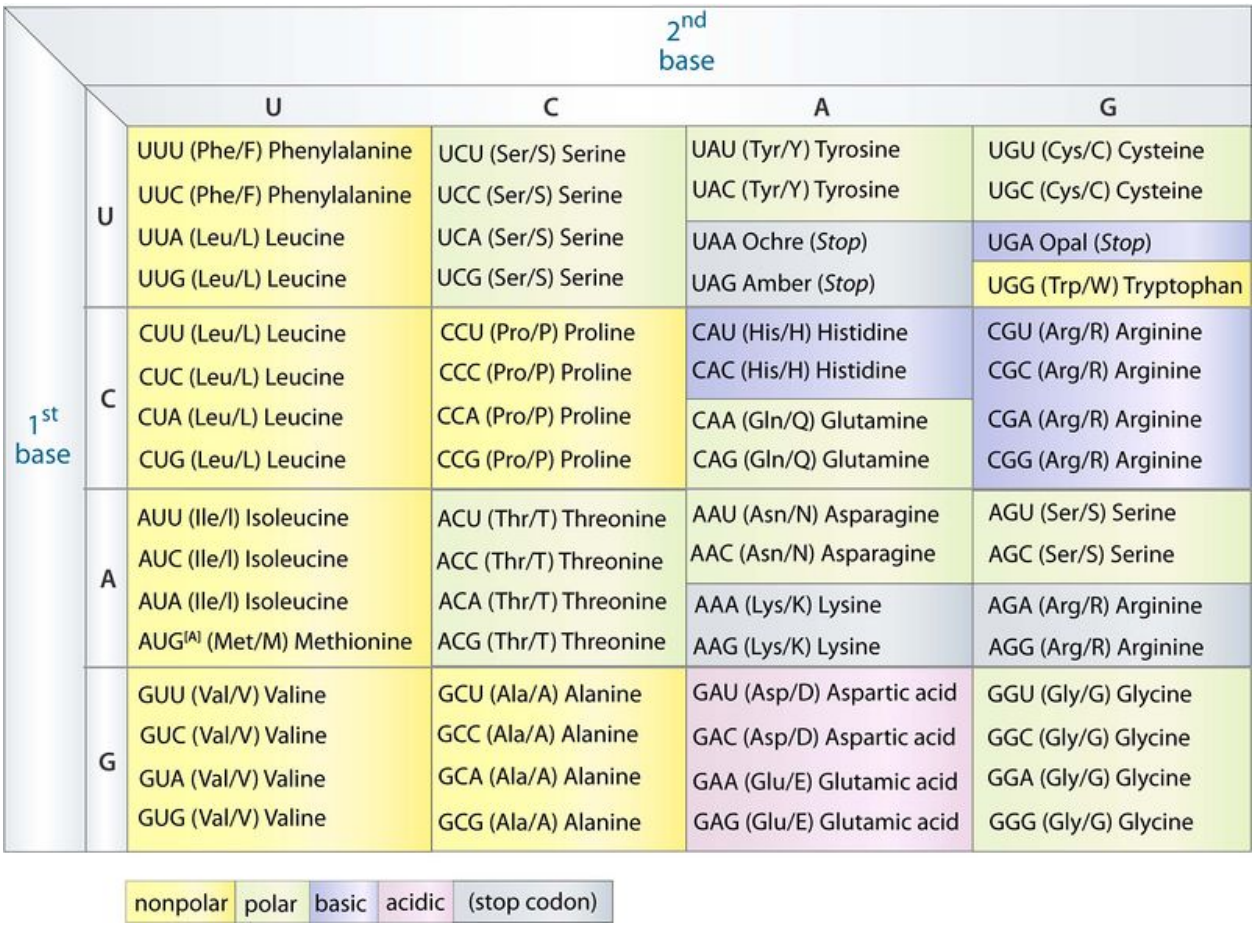
Splicing. Splicing removes introns from mRNA. UTR is an untranslated region of the mRNA.

The Genetic Code

How is the information in a gene encoded? The answer is the genetic code. The **genetic code** consists of the sequence of nitrogen bases—A, C, G, T (or U)—in a polynucleotide chain. The four bases make up the "letters" of the genetic code. The letters are combined in groups of three to form code "words," called **codons**. Each codon stands for (encodes) one amino acid, unless it codes for a start or stop signal. There are 20 common amino acids in proteins. There are 64 possible codons, more than enough to code for the 20 amino acids. The genetic code is shown in **Figure** 7.7. To see how scientists cracked the genetic code, go to this link: http://www.dnalc.org/view/16494-Animation-22-DNA-words-are-three-letters-long-.html.

Reading the Genetic Code

As shown in **Figure** 7.7, the codon AUG codes for the amino acid methionine. This codon is also the start codon that begins translation. The start codon establishes the reading frame of mRNA. The reading frame is the way the letters are divided into codons. After the AUG start codon, the next three letters are read as the second codon. The next



1st base	2nd base: U	2nd base: C	2nd base: A	2nd base: G
U	UUU (Phe/F) Phenylalanine	UCU (Ser/S) Serine	UAU (Tyr/Y) Tyrosine	UGU (Cys/C) Cysteine
	UUC (Phe/F) Phenylalanine	UCC (Ser/S) Serine	UAC (Tyr/Y) Tyrosine	UGC (Cys/C) Cysteine
	UUA (Leu/L) Leucine	UCA (Ser/S) Serine	UAA Ochre (*Stop*)	UGA Opal (*Stop*)
	UUG (Leu/L) Leucine	UCG (Ser/S) Serine	UAG Amber (*Stop*)	UGG (Trp/W) Tryptophan
C	CUU (Leu/L) Leucine	CCU (Pro/P) Proline	CAU (His/H) Histidine	CGU (Arg/R) Arginine
	CUC (Leu/L) Leucine	CCC (Pro/P) Proline	CAC (His/H) Histidine	CGC (Arg/R) Arginine
	CUA (Leu/L) Leucine	CCA (Pro/P) Proline	CAA (Gln/Q) Glutamine	CGA (Arg/R) Arginine
	CUG (Leu/L) Leucine	CCG (Pro/P) Proline	CAG (Gln/Q) Glutamine	CGG (Arg/R) Arginine
A	AUU (Ile/I) Isoleucine	ACU (Thr/T) Threonine	AAU (Asn/N) Asparagine	AGU (Ser/S) Serine
	AUC (Ile/I) Isoleucine	ACC (Thr/T) Threonine	AAC (Asn/N) Asparagine	AGC (Ser/S) Serine
	AUA (Ile/I) Isoleucine	ACA (Thr/T) Threonine	AAA (Lys/K) Lysine	AGA (Arg/R) Arginine
	AUG[A] (Met/M) Methionine	ACG (Thr/T) Threonine	AAG (Lys/K) Lysine	AGG (Arg/R) Arginine
G	GUU (Val/V) Valine	GCU (Ala/A) Alanine	GAU (Asp/D) Aspartic acid	GGU (Gly/G) Glycine
	GUC (Val/V) Valine	GCC (Ala/A) Alanine	GAC (Asp/D) Aspartic acid	GGC (Gly/G) Glycine
	GUA (Val/V) Valine	GCA (Ala/A) Alanine	GAA (Glu/E) Glutamic acid	GGA (Gly/G) Glycine
	GUG (Val/V) Valine	GCG (Ala/A) Alanine	GAG (Glu/E) Glutamic acid	GGG (Gly/G) Glycine

FIGURE 7.7

The Genetic Code. To find the amino acid for a particular codon, find the cell in the table for the first and second bases of the codon. Then, within that cell, find the codon with the correct third base. For example CUG codes for leucine, AAG codes for lysine, and GGG codes for glycine.

three letters after that are read as the third codon, and so on. This is illustrated in **Figure** 7.8. The mRNA molecule is read, codon by codon, until a stop codon is reached. UAG, UGA, and UAA are all stop codons. They do not code for any amino acids.

Characteristics of the Genetic Code

The genetic code has a number of important characteristics.

- The genetic code is universal. All known living things have the same genetic code. This shows that all organisms share a common evolutionary history.
- The genetic code is unambiguous. Each codon codes for just one amino acid (or start or stop). What might happen if codons encoded more than one amino acid?
- The genetic code is redundant. Most amino acids are encoded by more than one codon. In **Figure** 7.7, how many codons code for the amino acid threonine? What might be an advantage of having more than one codon

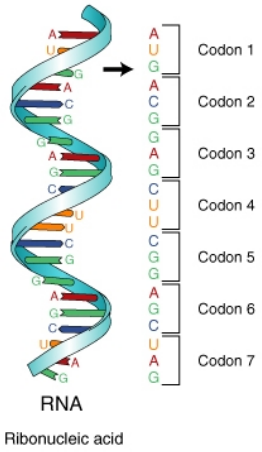


FIGURE 7.8

Reading the Genetic Code. The genetic code is read three bases at a time. Codons are the code words of the genetic code. Which amino acid does codon 2 in the drawing stand for?

for the same amino acid?

Translation

Translation is the second part of the central dogma of molecular biology: **RNA** $\rightarrow$ **Protein**. It is the process in which the genetic code in mRNA is read to make a protein. **Figure** 7.9 shows how this happens. After mRNA leaves the nucleus, it moves to a ribosome, which consists of rRNA and proteins. The ribosome reads the sequence of codons in mRNA. Molecules of tRNA bring amino acids to the ribosome in the correct sequence. To understand the role of tRNA, you need to know more about its structure. Each tRNA molecule has an anticodon for the amino acid it carries. An anticodon is complementary to the codon for an amino acid. For example, the amino acid lysine has the codon AAG, so the anticodon is UUC. Therefore, lysine would be carried by a tRNA molecule with the anticodon UUC. Wherever the codon AAG appears in mRNA, a UUC anticodon of tRNA temporarily binds. While bound to mRNA, tRNA gives up its amino acid. Bonds form between the amino acids as they are brought one by one to the ribosome, forming a polypeptide chain. The chain of amino acids keeps growing until a stop codon is reached. To see how this happens, go the link below. http://www.youtube.com/watch?v=B6O6uRb1D38&feature=related (1:29)

After a polypeptide chain is synthesized, it may undergo additional processes. For example, it may assume a folded shape due to interactions among its amino acids. It may also bind with other polypeptides or with different types of molecules, such as lipids or carbohydrates. Many proteins travel to the Golgi apparatus to be modified for the specific job they will do. You can see how this occurs by watching the animation at this link: http://vcell.ndsu.edu/animations/proteinmodification/movie-flash.htm.

Lesson Summary

- Transcription is the *DNA* $\rightarrow$ *RNA* part of the central dogma of molecular biology. It occurs in the nucleus. During transcription, a copy of mRNA is made that is complementary to a strand of DNA. In eukaryotes, mRNA may be modified before it leaves the nucleus.
- The genetic code consists of the sequence of bases in DNA or RNA. Groups of three bases form codons, and each codon stands for one amino acid (or start or stop). The codons are read in sequence following the start

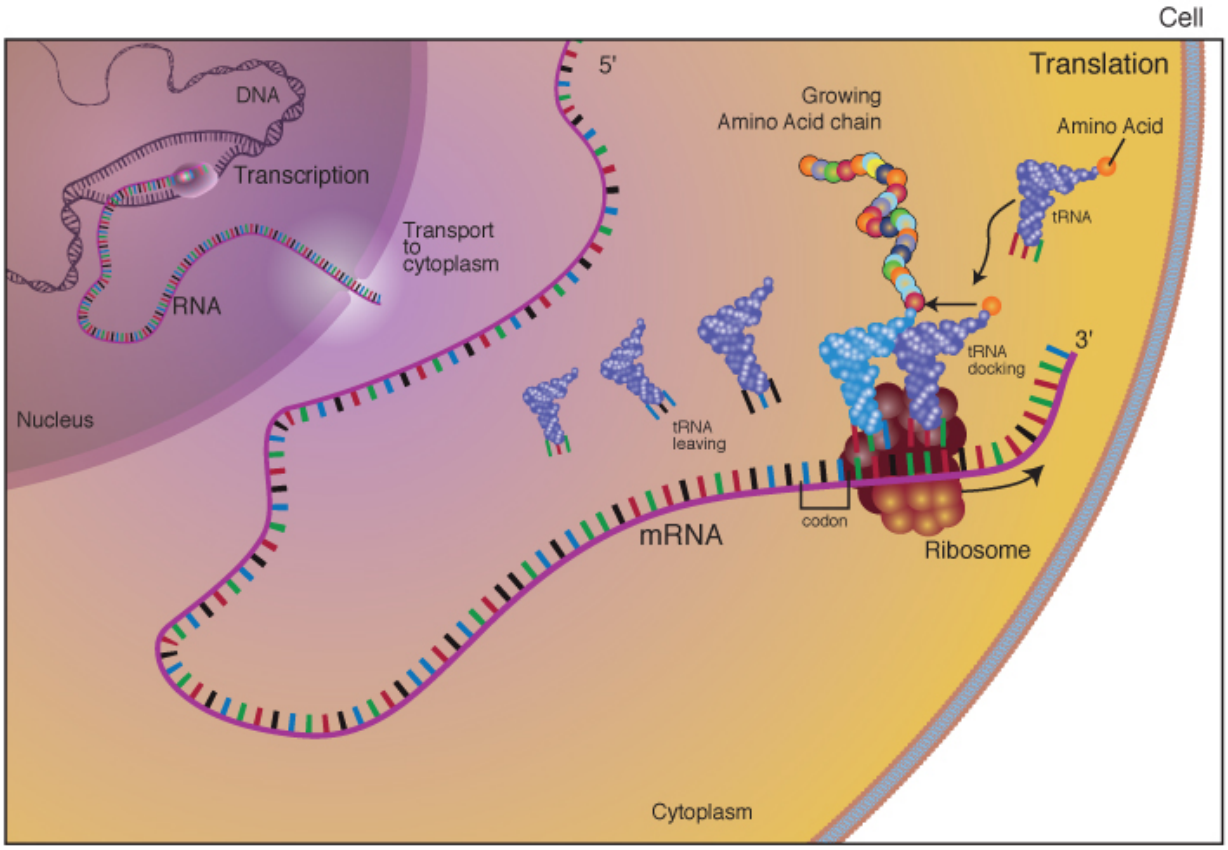


FIGURE 7.9

Translation. Translation of the codons in mRNA to a chain of amino acids occurs at a ribosome. Find the different types of RNA in the diagram. What are their roles in translation?

codon until a stop codon is reached. The genetic code is universal, unambiguous, and redundant.

- Translation is the *RNA* $\rightarrow$ *protein* part of the central dogma. It occurs at a ribosome. During translation, a protein is synthesized using the codons in mRNA as a guide. All three types of RNA play a role in translation.

Lesson Review Questions

Recall

1. Describe transcription.
2. How may mRNA be modified before it leaves the nucleus?
3. What is the genetic code? What are codons?
4. Outline the steps of translation.

Apply Concepts

5. Use the genetic code in **Figure** 7.7 to translate the following segment of RNA into a sequence of five amino acids:
GUC-GCG-CAU-AGC-AAG

Think Critically

6. The genetic code is universal, unambiguous, and redundant. Explain what this means and why it is important.
7. How are transcription and translation related to the central dogma of molecular biology?

Points to Consider

When DNA is replicated or transcribed, accidents can happen, leading to a change in the base sequence.

- What do you think could cause such accidents to occur?
- How might the changes affect the reading frame? How might the encoded protein be affected?

7.3 Mutation

Lesson Objectives

- Identify causes of mutation.
- Compare and contrast types of mutations.
- Explain how mutations may affect the organisms in which they occur.

Vocabulary

chromosomal alteration mutation that changes chromosome structure

frameshift mutation deletion or insertion of one or more nucleotides that changes the reading frame of the genetic material

genetic disorder disease caused by a mutation in one or a few genes

germline mutation mutation that occur in gametes

mutagen environmental factors that causes mutations

mutation change in the sequence of bases in DNA or RNA

point mutation change in a single nucleotide base in the genetic material

somatic mutation mutation that occurs in cells of the body other than gametes

Introduction

A change in the sequence of bases in DNA or RNA is called a **mutation**. Does the word mutation make you think of science fiction and bug-eyed monsters? Think again. Everyone has mutations. In fact, most people have dozens or even hundreds of mutations in their DNA. Mutations are essential for evolution to occur. They are the ultimate source of all new genetic material in a species. Although most mutations have no effect on the organisms in which they occur, some mutations are beneficial. Even harmful mutations rarely cause drastic changes in organisms.

Causes of Mutation

Mutations have many possible causes. Some mutations seem to happen spontaneously without any outside influence. They occur when mistakes are made during DNA replication or transcription. Other mutations are caused by environmental factors. Anything in the environment that can cause a mutation is known as a **mutagen**. Examples of mutagens are pictured in **Figure** 7.10. For a video about mutagens, go the link below. http://www.youtube.com/watch?v=0wrNxCGKCws&feature=related (0:36)

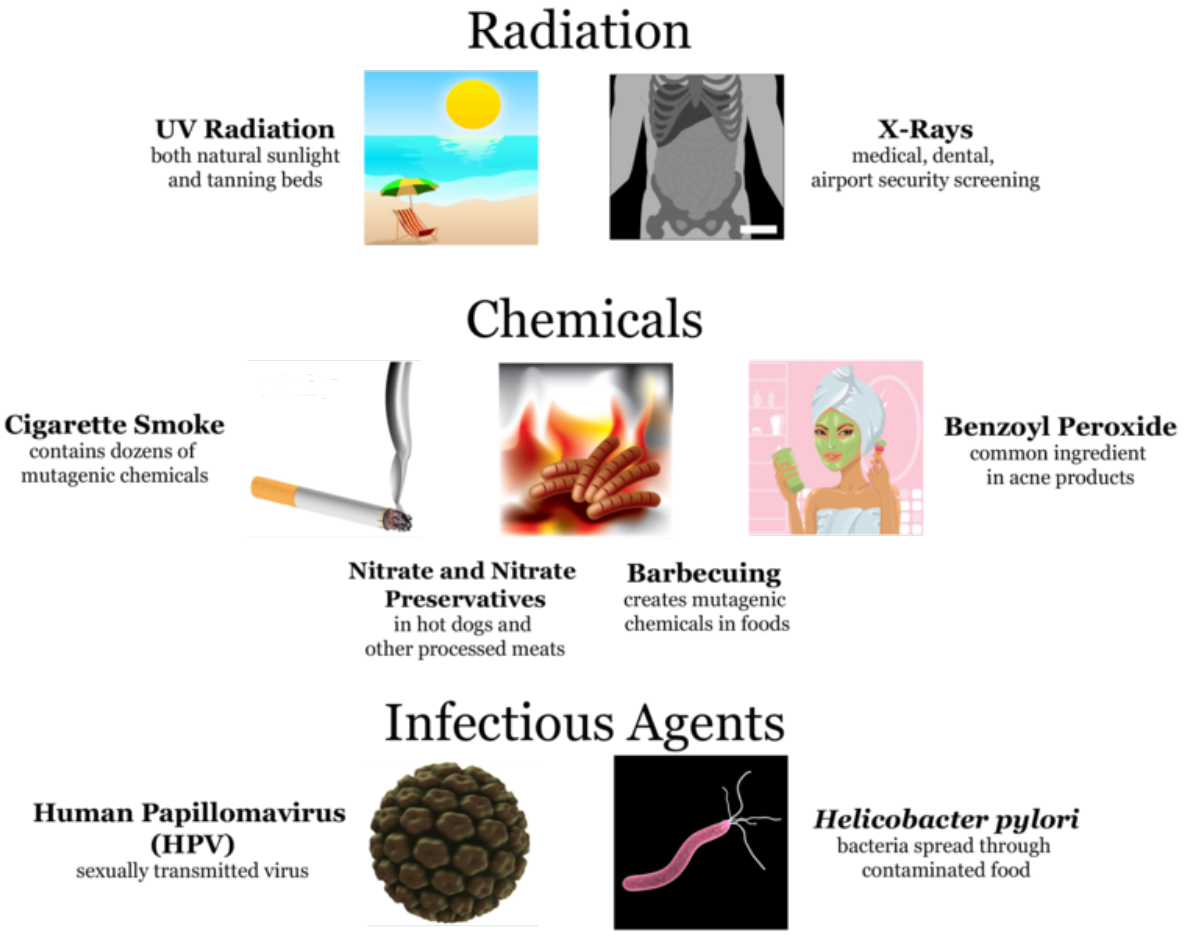


FIGURE 7.10

Examples of Mutagens. Types of mutagens include radiation, chemicals, and infectious agents. Do you know of other examples of each type of mutagen shown here?

Types of Mutations

There are a variety of types of mutations. Two major categories of mutations are germline mutations and somatic mutations.

- **Germline mutations** occur in gametes. These mutations are especially significant because they can be transmitted to offspring and every cell in the offspring will have the mutation.
- **Somatic mutations** occur in other cells of the body. These mutations may have little effect on the organism because they are confined to just one cell and its daughter cells. Somatic mutations cannot be passed on to offspring.

Mutations also differ in the way that the genetic material is changed. Mutations may change the structure of a chromosome or just change a single nucleotide.

Chromosomal Alterations

Chromosomal alterations are mutations that change chromosome structure. They occur when a section of a chromosome breaks off and rejoins incorrectly or does not rejoin at all. Possible ways these mutations can occur are illustrated in **Figure** 7.11. Go to this link for a video about chromosomal alterations: http://www.youtube.com/watch?v=OrXRSqa_3lU&feature=related (2:18).

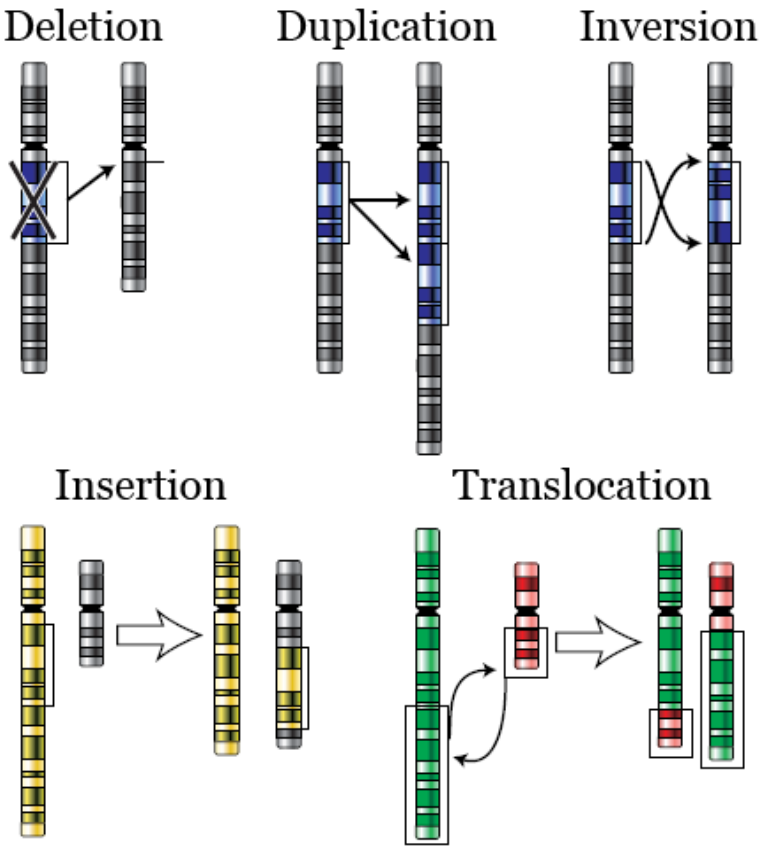


FIGURE 7.11

Chromosomal Alterations. Chromosomal alterations are major changes in the genetic material.

Chromosomal alterations are very serious. They often result in the death of the organism in which they occur. If

the organism survives, it may be affected in multiple ways. An example of a human chromosomal alteration is the mutation that causes Down Syndrome. It is a duplication mutation that leads to developmental delays and other abnormalities.

Point Mutations

A **point mutation** is a change in a single nucleotide in DNA. This type of mutation is usually less serious than a chromosomal alteration. An example of a point mutation is a mutation that changes the codon UUU to the codon UCU. Point mutations can be silent, missense, or nonsense mutations, as shown in **Table** 7.1. The effects of point mutations depend on how they change the genetic code. You can watch an animation about nonsense mutations at this link: http://www.biostudio.com/d_%20Nonsense%20Suppression%20I%20Nonsense%20Mutation.htm.

TABLE 7.1: Point Mutations and Their Effects

Type	Description	Example	Effect
Silent	mutated codon codes for the same amino acid	CAA (glutamine) → CAG (glutamine)	none
Missense	mutated codon codes for a different amino acid	CAA (glutamine) → CCA (proline)	variable
Nonsense	mutated codon is a premature stop codon	CAA (glutamine) → UAA (stop) usually	serious

Frameshift Mutations

A **frameshift mutation** is a deletion or insertion of one or more nucleotides that changes the reading frame of the base sequence. Deletions remove nucleotides, and insertions add nucleotides. Consider the following sequence of bases in RNA:

AUG-AAU-ACG-GCU = start-asparagine-threonine-alanine

Now, assume an insertion occurs in this sequence. Let's say an **A** nucleotide is inserted after the start codon **AUG**:

AUG-AAA-UAC-GGC-U = start-lysine-tyrosine-glycine

Even though the rest of the sequence is unchanged, this insertion changes the reading frame and thus all of the codons that follow it. As this example shows, a frameshift mutation can dramatically change how the codons in mRNA are read. This can have a drastic effect on the protein product.

Effects of Mutations

The majority of mutations have neither negative nor positive effects on the organism in which they occur. These mutations are called neutral mutations. Examples include silent point mutations. They are neutral because they do not change the amino acids in the proteins they encode. Many other mutations have no effect on the organism because they are repaired before protein synthesis occurs. Cells have multiple repair mechanisms to fix mutations in DNA. One way DNA can be repaired is illustrated in **Figure** 7.12. If a cell's DNA is permanently damaged and cannot be repaired, the cell is likely to be prevented from dividing.

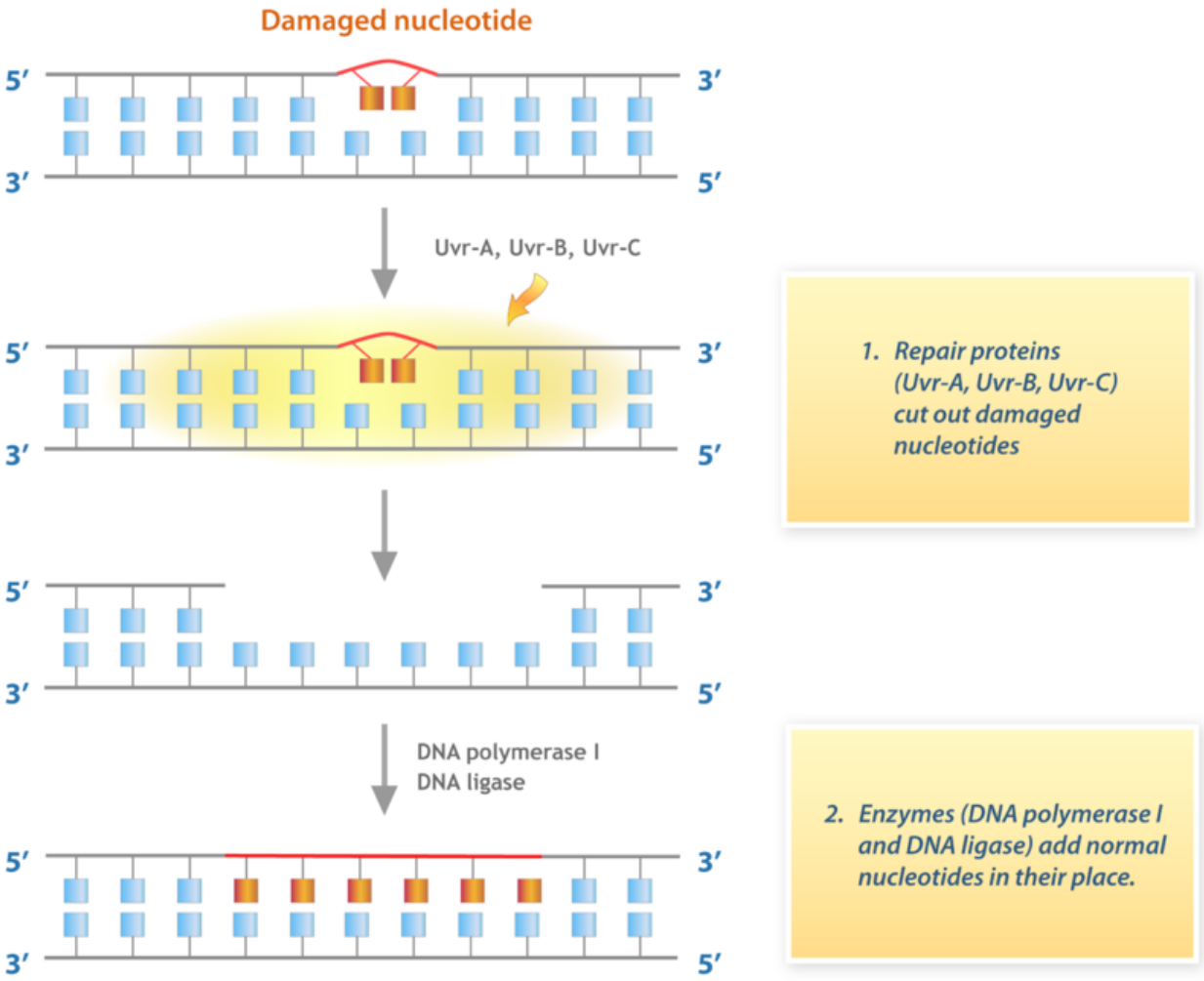


FIGURE 7.12

DNA Repair Pathway. This flow chart shows one way that damaged DNA is repaired in E. coli bacteria.

Beneficial Mutations

Some mutations have a positive effect on the organism in which they occur. They are called beneficial mutations. They lead to new versions of proteins that help organisms adapt to changes in their environment. Beneficial mutations are essential for evolution to occur. They increase an organism's changes of surviving or reproducing, so they are likely to become more common over time. There are several well-known examples of beneficial mutations. Here are just two:

a. Mutations in many bacteria that allow them to survive in the presence of antibiotic drugs. The mutations lead to antibiotic-resistant strains of bacteria.
b. A unique mutation is found in people in a small town in Italy. The mutation protects them from developing atherosclerosis, which is the dangerous buildup of fatty materials in blood vessels. The individual in which the mutation first appeared has even been identified.

Harmful Mutations

Imagine making a random change in a complicated machine such as a car engine. The chance that the random change would improve the functioning of the car is very small. The change is far more likely to result in a car that does not run well or perhaps does not run at all. By the same token, any random change in a gene's DNA is likely to result in a protein that does not function normally or may not function at all. Such mutations are likely to be harmful. Harmful mutations may cause genetic disorders or cancer.

- A **genetic disorder** is a disease caused by a mutation in one or a few genes. A human example is cystic fibrosis. A mutation in a single gene causes the body to produce thick, sticky mucus that clogs the lungs and blocks ducts in digestive organs. You can watch a video about cystic fibrosis and other genetic disorders at this link: http://www.youtube.com/watch?v=8s4he3wLgkM&feature=PlayList&p=397710758E9BCB24&playnext_from=PL&playnext=1&index=17 (9:31).

- Cancer is a disease in which cells grow out of control and form abnormal masses of cells. It is generally caused by mutations in genes that regulate the cell cycle. Because of the mutations, cells with damaged DNA are allowed to divide without limits. Cancer genes can be inherited. You can learn more about hereditary cancer by watching the video at the following link: http://www.youtube.com/watch?v=LWk5FplsKwM (4:29)

Lesson Summary

- Mutations are caused by environmental factors known as mutagens. Types of mutagens include radiation, chemicals, and infectious agents.
- Germline mutations occur in gametes. Somatic mutations occur in other body cells. Chromosomal alterations are mutations that change chromosome structure. Point mutations change a single nucleotide. Frameshift mutations are additions or deletions of nucleotides that cause a shift in the reading frame.
- Mutations are essential for evolution to occur because they increase genetic variation and the potential for individuals to differ. The majority of mutations are neutral in their effects on the organisms in which they occur. Beneficial mutations may become more common through natural selection. Harmful mutations may cause genetic disorders or cancer.

Lesson Review Questions

Recall

1. Define mutation and mutagen.
2. List three examples of mutagens.
3. Identify three types of chromosomal alterations.
4. Distinguish among silent, missense, and nonsense point mutations.
5. What is a frameshift mutation? What causes this type of mutation?

Apply Concepts

6. Assume that a point mutation changes the codon AUU to AUC. Why is this a neutral mutation?

7. Look at the mutation shown below. The base A was inserted following the start codon AUG. Describe how this mutation affects the encoded amino acid sequence.

AUG-GUC-CCU-AAA → AUG-AGU-CCC-UAA-A

Think Critically

8. Compare and contrast germline mutations and somatic mutations.

9. Why are mutations essential for evolution to occur?

Points to Consider

Sometimes even drastic mutations do not affect the proteins produced by a particular type of cell. The reason? The genes affected by the mutations are not normally used to make proteins in that type of cell. In all cells, some genes are turned off - they are not transcribed - while other genes are turned on.

- How do cells control which genes are turned on and used to make proteins?
- Can you think of a mechanism that might prevent transcription of a gene?

7.4 Regulation of Gene Expression

Lesson Objectives

- Identify general mechanisms that regulate gene expression.
- Describe how gene regulation occurs in prokaryotes.
- Give an overview of gene regulation in eukaryotes.

Vocabulary

gene expression use of a gene to make a protein

homeobox gene gene that codes of regulatory proteins that control gene expression during development

operator a region of an operon where regulatory proteins bind

operon region of prokaryotic DNA that consists of a promoter, an operator, and one or more genes that encode proteins needed for a specific function

regulatory element region of DNA where a regulatory protein binds

regulatory protein protein that regulates gene expression

TATA box regulatory element that is part of the promoter of most eukaryotic genes

Introduction

Each of your cells has at least 20,000 genes. In fact, all of your cells have the same genes. Do all of your cells make the same proteins? Obviously not. If they did, then all your cells would be alike. Instead, you have cells with different structures and functions. This is because different cells make different proteins. They do this by using, or expressing, different genes. Using a gene to make a protein is called **gene expression**.

How Gene Expression is Regulated

Gene expression is regulated to ensure that the correct proteins are made when and where they are needed. Regulation may occur at any point in the expression of a gene, from the start of transcription to the processing of a protein after translation. The focus in this lesson is the regulation of transcription. As shown in **Figure** 7.13, transcription

is controlled by **regulatory proteins**. The proteins bind to regions of DNA, called **regulatory elements**, which are located near promoters. After regulatory proteins bind to regulatory elements, they can interact with RNA polymerase, the enzyme that transcribes DNA to mRNA. Regulatory proteins are typically either activators or repressors.

- Activators promote transcription by enhancing the interaction of RNA polymerase with the promoter.
- Repressors prevent transcription by impeding the progress of RNA polymerase along the DNA strand.

Other factors may also be involved in the regulation of transcription, but these are typically the key players.

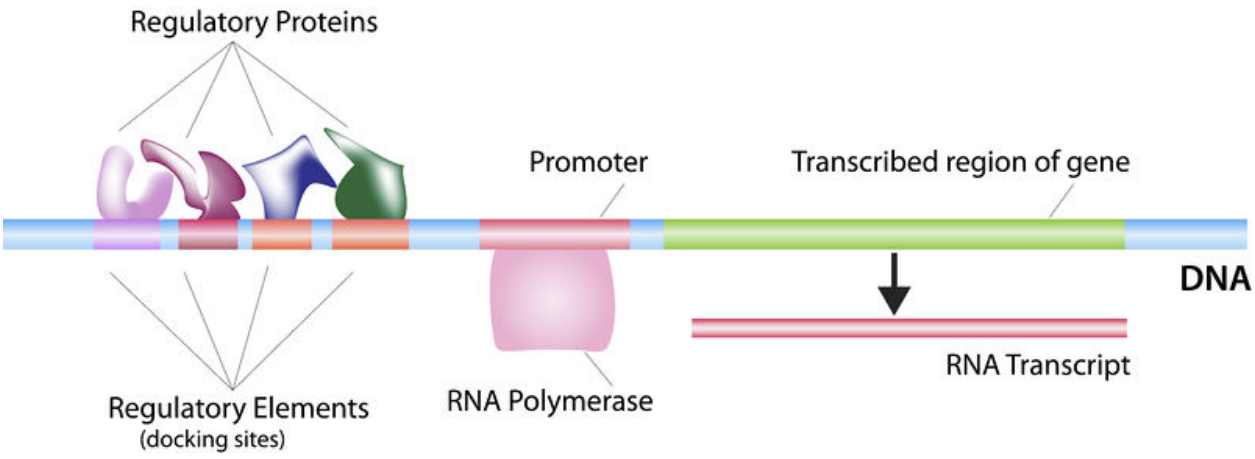


FIGURE 7.13

Regulation of Transcription. Regulatory proteins bind to regulatory elements to control transcription. The regulatory elements are embedded within the DNA.

Prokaryotic Gene Regulation

Transcription is regulated differently in prokaryotes and eukaryotes. In general, prokaryotic regulation is simpler than eukaryotic regulation.

The Role of Operons

Regulation of transcription in prokaryotes typically involves operons. An **operon** is a region of DNA that consists of one or more genes that encode the proteins needed for a specific function. The operon also includes a promoter and an operator. The **operator** is a region of the operon where regulatory proteins bind. It is located near the promoter and helps regulate transcription of the operon genes.

The Lac Operon

A well-known example of operon regulation involves the lac operon in *E. coli* bacteria (see **Figure** 7.14 and the video at the link below). The lac operon consists of a promoter, an operator, and three genes that encode the enzymes

needed to digest lactose, the sugar found in milk. The lac operon is regulated by lactose in the environment. http://www.youtube.com/watch?v=oBwtxdI1zvk

- When lactose is absent, a repressor protein binds to the operator. The protein blocks the binding of RNA polymerase to the promoter. As a result, the lac genes are not expressed.
- When lactose is present, the repressor protein does not bind to the operator. This allows RNA polymerase to bind to the promoter and begin transcription. As a result, the lac genes are expressed, and lactose is digested.

Why might it be beneficial to express genes only when they are needed? (Hint: synthesizing proteins requires energy and materials.)

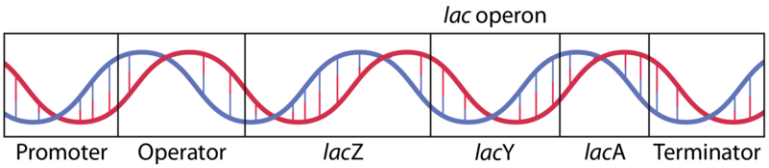


FIGURE 7.14

The three genes of the lac operon are lacZ, lacY, and lacA. They encode proteins needed to digest lactose. The genes are expressed only in the presence of lactose.

Eukaryotic Gene Regulation

In eukaryotic cells, the start of transcription is one of the most complicated parts of gene regulation. There may be many regulatory proteins and regulatory elements involved. Regulation may also involve enhancers. Enhancers are distant regions of DNA that can loop back to interact with a gene's promoter.

The TATA Box

Different types of cells have unique patterns of regulatory elements that result in only the necessary genes being transcribed. That's why a skin cell and nerve cell, for example, are so different from each other. However, some patterns of regulatory elements are common to all genes, regardless of the cells in which they occur. An example is the **TATA box**. This is a regulatory element that is part of the promoter of most eukaryotic genes. A number of regulatory proteins bind to the TATA box, forming a multi-protein complex. It is only when all of the appropriate proteins are bound to the TATA box that RNA polymerase recognizes the complex and binds to the promoter. Once RNA polymerase binds, transcription begins. To see a video showing the role of the TATA box in the initiation of transcription, go to this link: http://www.youtube.com/watch?v=6tqPsI-9aQA&feature=related.

Regulation During Development

The regulation of gene expression is extremely important during the development of an organism. Regulatory proteins must turn on certain genes in particular cells at just the right time so the organism develops normal organs and organ systems. **Homeobox genes** are an example of genes that regulate development. They code for regulatory proteins that switch on whole series of major developmental genes. In insects, homeobox genes called hox genes ensure that body parts such as limbs develop in the correct place. **Figure** 7.15 shows how a mutation in a hox gene can affect an insect's development. You can learn more about homeobox genes at this link: http://www.youtube.com/watch?v=LFG-aLidT8s.

FIGURE 7.15

Effect of Hox Gene Mutation. Scientists caused a mutation in a hox gene of this fruit fly. As a result of the mutation, a leg grew out of its head where an antenna should have developed.

Gene Expression and Cancer

The mutations that cause cancer generally occur in two types of regulatory genes: tumor-suppressor genes and proto-oncogenes (see **Figure** 7.16). These genes produce regulatory proteins that control the cell cycle. When the genes mutate, cells with mutations divide rapidly and without limits.

Lesson Summary

- Gene transcription is controlled by regulatory proteins that bind to regulatory elements on DNA. The proteins usually either activate or repress transcription.
- Regulation of transcription in prokaryotes typically involves an operon, such as the lac operon in *E. coli*. The lac operon is regulated by proteins that behave differently depending on whether lactose is present.
- Regulation of transcription in eukaryotes is generally more complex. It involves unique regulatory elements in different cells as well as common regulatory elements such as the TATA box. Regulation is especially important during development. It may involve regulatory genes such as homeobox genes that switch other regulatory genes on or off. Mutations in regulatory genes that normally control the cell cycle cause cancer.

Lesson Review Questions

Recall

1. What is gene expression?
2. Describe how regulatory proteins regulate gene expression.
3. Identify the TATA box and its function in transcription.

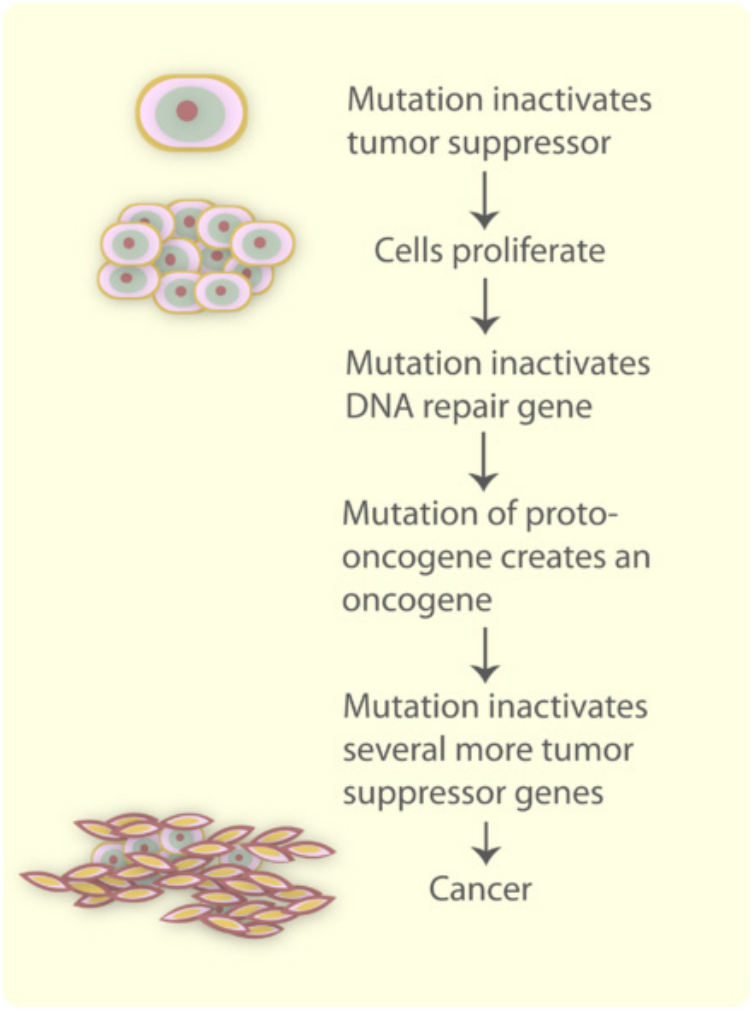


FIGURE 7.16

How Cancer Develops. This flow chart shows how a series of mutations in tumor-suppressor genes and proto-oncogenes leads to cancer.

4. What is a homeobox gene?

Apply Concepts

5. Draw a diagram to show how the lac operon is regulated.
6. Sketch how an insect with a mutated hox gene might look. Explain your sketch.

Think Critically

7. Why is gene regulation especially important during development?

Points to Consider

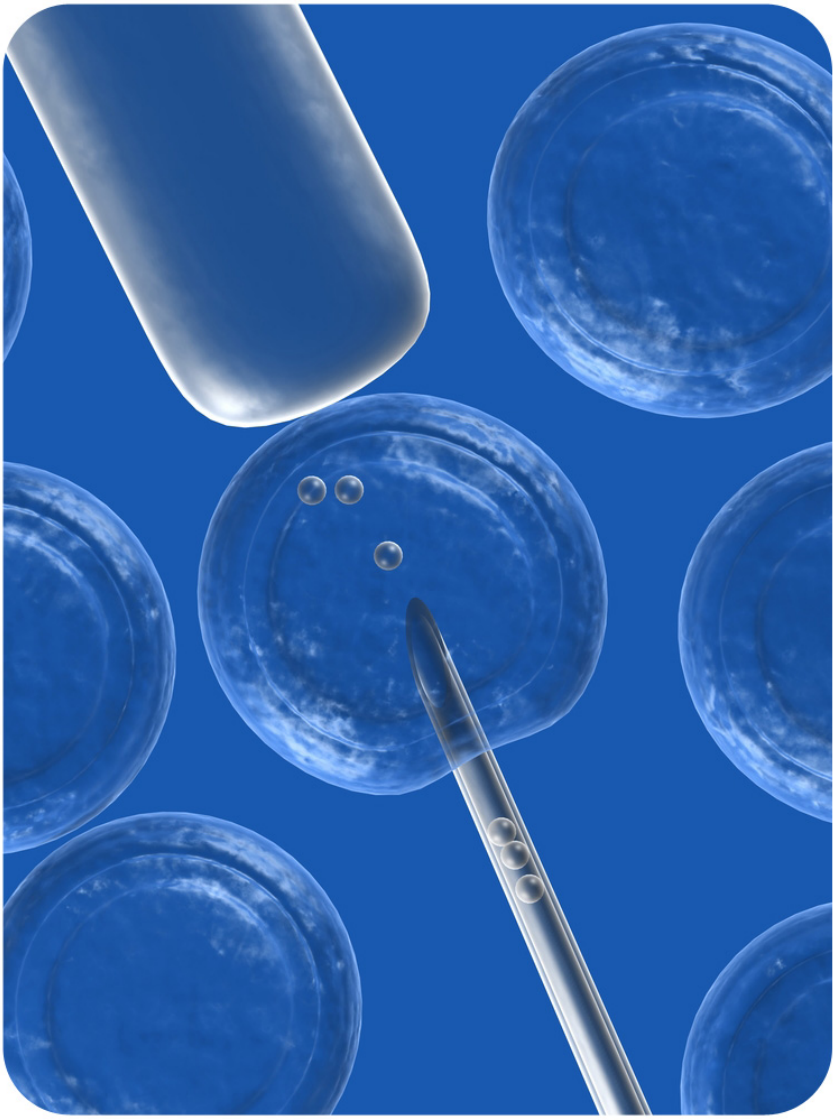
Scientists know more about human chromosomes and genes than they know about the genetic material of most other species. In fact, scientists have identified all of the approximately 20,000-25,000 genes in human DNA.

- What do you know about human chromosomes and genes? For example, do you know how many chromosomes humans normally have?
- Do you know how human characteristics are inherited? Can you identify characteristics that are controlled by a single gene?

CHAPTER 8

Human Genetics and Biotechnology

Chapter Outline



Biotechnology. Gene Therapy. Reality or fiction? During your lifetime, gene therapy may be mainstream medicine. Here we see a representation of the insertion of DNA into the nucleus of a cell. Is this possible? Yes. In this chapter, you will learn how human chromosomes and genes are inherited and how they control the traits that make each of us unique, how they can cause disease, and how those diseases can be treated.

8.1 Human Chromosomes and Genes

Lesson Objective

- Define the human genome.
- Describe human chromosomes and genes.
- Explain linkage and linkage maps.

Vocabulary

autosome chromosomes 1–22 in humans that contain genes for characteristics unrelated to sex

human genome all of the DNA of the human species

Human Genome Project international science project that sequenced all 3 billion base pairs of the human genome

linkage map map that shows the positions of genes on a chromosome based on the frequency of crossing-over between the genes

linked genes genes that are located on the same chromosome

sex chromosome X or Y chromosome (in humans)

sex-linked gene gene located on a sex chromosome

X-linked gene gene located on the X chromosome

Introduction

Nobody else in the world is exactly like you. What makes you different from everyone else? Genes have a lot to do with it. Unless you have an identical twin, no one else on Earth has exactly the same genes as you. What about identical twins? Are they identical in every way? They develop from the same fertilized egg, so they have all same genes, but even they are not completely identical. Why? The environment also influences human characteristics, and no two people have exactly the same environment.

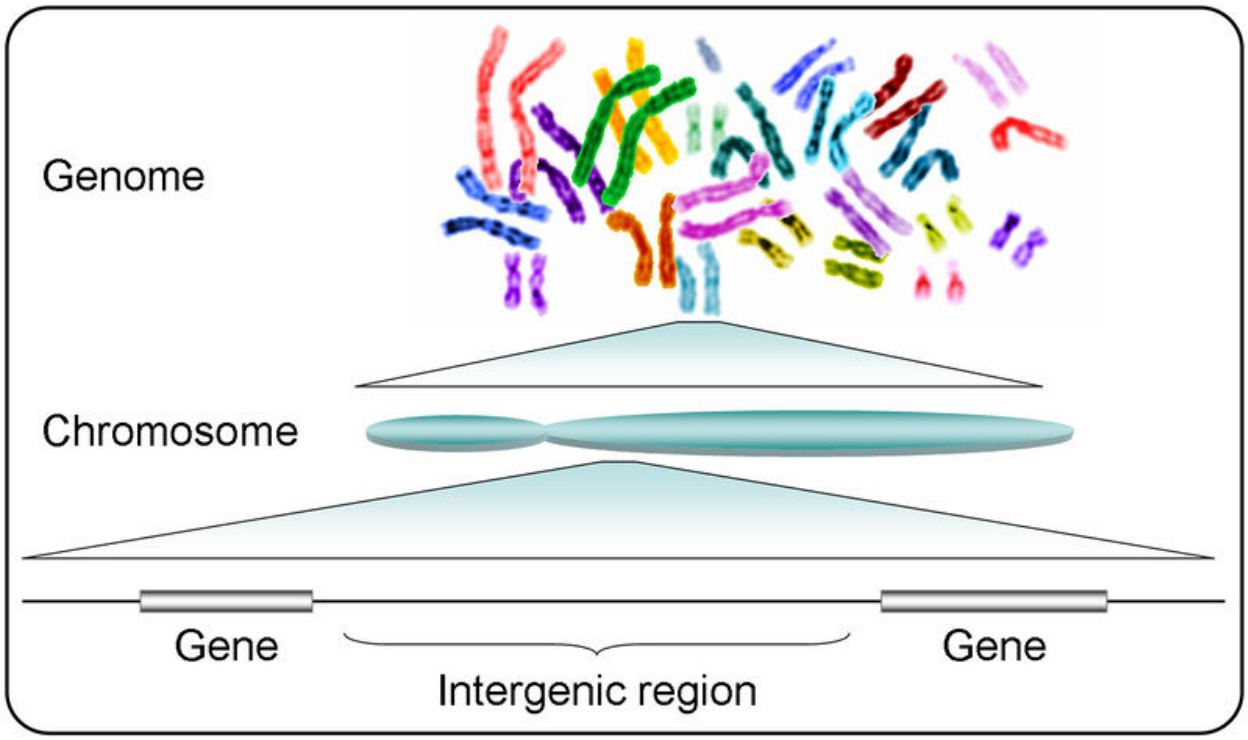


FIGURE 8.1

Human Genome, Chromosomes, and Genes. Each chromosome of the human genome contains many genes as well as noncoding intergenic (between genes) regions. Each pair of chromosomes is shown here in a different color.

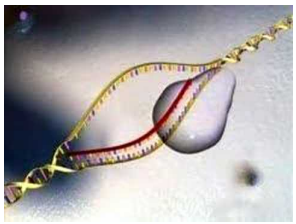
The Human Genome

All the DNA of the human species makes up the **human genome**. This DNA consists of about 3 billion base pairs and is divided into thousands of genes on 23 pairs of chromosomes. The human genome also includes noncoding sequences of DNA, as shown in **Figure** 8.1.

Thanks to the **Human Genome Project**, scientists now know the DNA sequence of the entire human genome. The Human Genome Project is an international project that includes scientists from around the world. It began in 1990, and by 2003, scientists had sequenced all 3 billion base pairs of human DNA. Now they are trying to identify all the genes in the sequence.

You can watch a video about the Human Genome Project and how it cracked the *code of life* at this link: http://www.pbs.org/wgbh/nova/genome/program.html.

Our Molecular Selves video discusses the human genome, and is available at http://www.genome.gov/25520211 or http://www.youtube.com/watch?v=XuUpnAz5y1g&feature=related.



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Chromosomes and Genes

Each species has a characteristic number of chromosomes. The human species is characterized by 23 pairs of chromosomes, as shown in **Figure** 8.2 and **Figure** 8.3. You can watch a short animation about human chromosomes at this link: http://www.dnalc.org/view/15520-DNA-is-organized-into-46-chromosomes-including-sex-chromosomes-3D-animation.html.

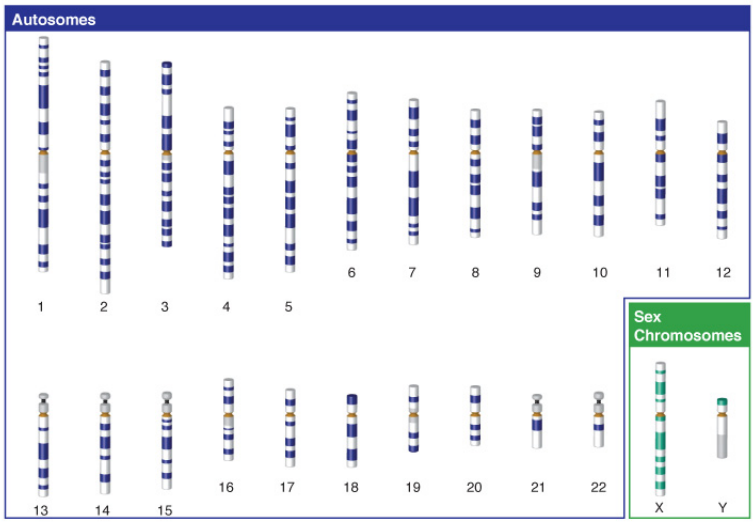


FIGURE 8.2

Human Chromosomes. Human chromosomes are shown here arranged by size. Chromosome 1 is the largest, and chromosome 22 is the smallest. All normal human cells (except gametes) have two of each chromosome, for a total of 46 chromosomes per cell. Only one of each pair is shown here.

Autosomes

Of the 23 pairs of human chromosomes, 22 pairs are autosomes (numbers 1–22 in **Figure** 8.3). **Autosomes** are chromosomes that contain genes for characteristics that are unrelated to sex. These chromosomes are the same in males and females. The great majority of human genes are located on autosomes. At the link below, you can click on any human chromosome to see which traits its genes control. http://www.ornl.gov/sci/techresources/Human_Genome/posters/chromosome/chooser.shtml

Sex Chromosomes

The remaining pair of human chromosomes consists of the **sex chromosomes**, X and Y. Females have two X chromosomes, and males have one X and one Y chromosome. In females, one of the X chromosomes in each cell is inactivated and known as a Barr body. This ensures that females, like males, have only one functioning copy of the X chromosome in each cell. As you can see from **Figure** 8.2 and **Figure** 8.3, the X chromosome is much larger than the Y chromosome. The X chromosome has about 2,000 genes, whereas the Y chromosome has fewer

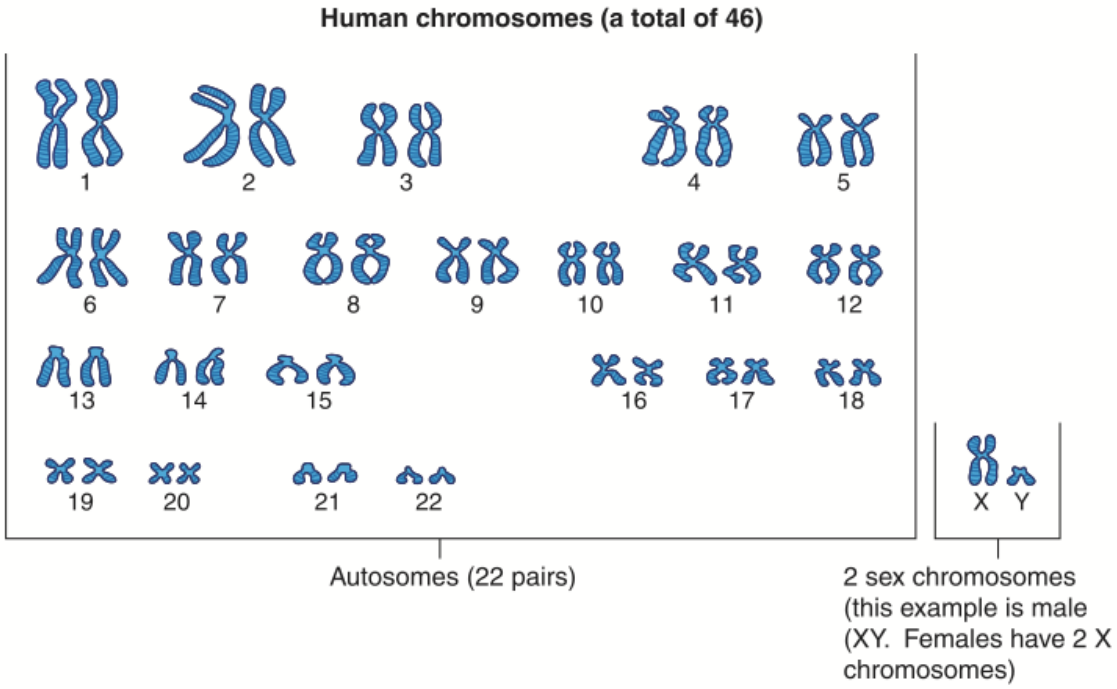


FIGURE 8.3

Human Chromosomes. Humans have 23 pairs of chromosomes. Pairs 1-22 are autosomes. Females have two X chromosomes, and males have an X and a Y chromosome.

than 100, none of which are essential to survival. Virtually all of the X chromosome genes are unrelated to sex. Only the Y chromosome contains genes that determine sex. A single Y chromosome gene, called SRY (which stands for sex-determining region Y gene), triggers an embryo to develop into a male. Without a Y chromosome, an individual develops into a female, so you can think of female as the default sex of the human species. Can you think of a reason why the Y chromosome is so much smaller than the X chromosome? At the link that follows, you can watch an animation that explains why: http://www.hhmi.org/biointeractive/gender/Y_evolution.html.

Human Genes

Humans have an estimated 20,000 to 22,000 genes. This may sound like a lot, but it really isn't. Far simpler species have almost as many genes as humans. However, human cells use splicing and other processes to make multiple proteins from the instructions encoded in a single gene. Of the 3 billion base pairs in the human genome, only about 25 percent make up genes and their regulatory elements. The functions of many of the other base pairs are still unclear. To learn more about the coding and noncoding sequences of human DNA, watch the animation at this link: http://www.hhmi.org/biointeractive/dna/DNAi_coding_sequences.html.

The majority of human genes have two or more possible alleles. Differences in alleles account for the considerable genetic variation among people. In fact, most human genetic variation is the result of differences in individual DNA bases within alleles.

Linkage

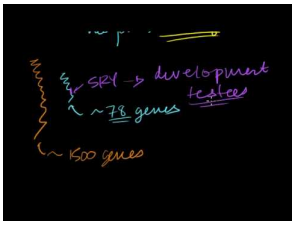
Genes that are located on the same chromosome are called **linked genes**. Alleles for these genes tend to segregate together during meiosis, unless they are separated by crossing-over. Crossing-over occurs when two homologous chromosomes exchange genetic material during meiosis I. The closer together two genes are on a chromosome, the less likely their alleles will be separated by crossing-over. At the following link, you can watch an animation showing how genes on the same chromosome may be separated by crossing-over: http://www.biostudio.com/d_%20Meiotic%20Recombination%20Between%20Linked%20Genes.htm.

Linkage explains why certain characteristics are frequently inherited together. For example, genes for hair color and eye color are linked, so certain hair and eye colors tend to be inherited together, such as blonde hair with blue eyes and brown hair with brown eyes. What other human traits seem to occur together? Do you think they might be controlled by linked genes?

Sex-Linked Genes

Genes located on the sex chromosomes are called **sex-linked genes**. Most sex-linked genes are on the X chromosome, because the Y chromosome has relatively few genes. Strictly speaking, genes on the X chromosome are **X-linked genes**, but the term sex-linked is often used to refer to them.

Sex-linked traits are discussed at http://www.youtube.com/user/khanacademy#p/c/7A9646BC5110CF64/15/-ROhfKyxgCo (14:19).



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Mapping Linkage

Linkage can be assessed by determining how often crossing-over occurs between two genes on the same chromosome. Genes on different (nonhomologous) chromosomes are not linked. They assort independently during meiosis, so they have a 50 percent chance of ending up in different gametes. If genes show up in different gametes less than 50 percent of the time (that is, they tend to be inherited together), they are assumed to be on the same (homologous) chromosome. They may be separated by crossing-over, but this is likely to occur less than 50 percent of the time. The lower the frequency of crossing-over, the closer together on the same chromosome the genes are presumed to be. Frequencies of crossing-over can be used to construct a linkage map like the one in **Figure** 8.4. A **linkage map** shows the locations of genes on a chromosome.

Lesson Summary

- The human genome consists of about 3 billion base pairs of DNA. In 2003, the Human Genome Project finished sequencing all 3 billion base pairs.
- Humans have 23 pairs of chromosomes. Of these, 22 pairs are autosomes. The X and Y chromosomes are the sex chromosomes. Females have two X chromosomes, and males have one X and one Y. Human chromosomes contain a total of 20,000 to 22,000 genes, the majority of which have two or more alleles.

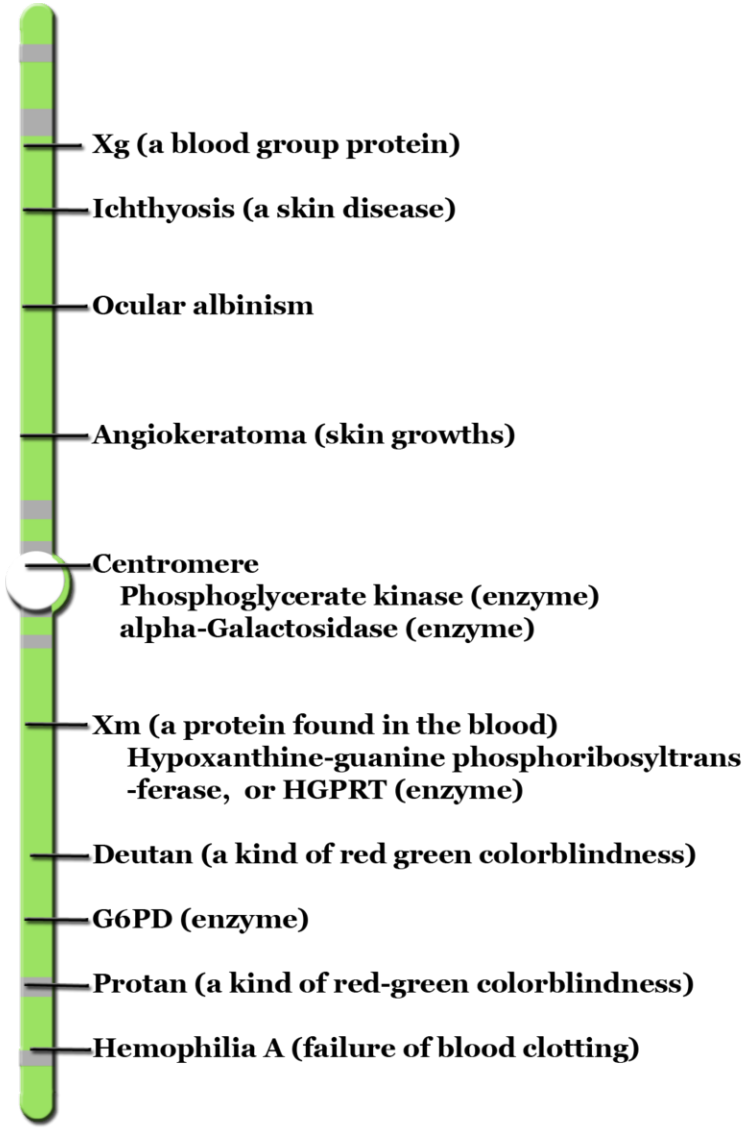


FIGURE 8.4

Linkage Map for the Human X Chromosome. This linkage map shows the locations of several genes on the X chromosome. Some of the genes code for normal proteins. Others code for abnormal proteins that lead to genetic disorders. Which pair of genes would you expect to have a lower frequency of crossing-over: the genes that code for hemophilia A and G6PD deficiency, or the genes that code for protan and Xm?

- Linked genes are located on the same chromosome. Sex-linked genes are located on a sex chromosome, and X-linked genes are located on the X chromosome. The frequency of crossing-over between genes is used to construct linkage maps that show the locations of genes on chromosomes.

Lesson Review Questions

Recall

1. Describe the human genome.
2. What has the Human Genome Project achieved?
3. What are linked genes?
4. Describe human genetic variation.

Apply Concepts

5. Explain how you would construct a linkage map for a human chromosome. What data would you need?

Think Critically

6. Compare and contrast human autosomes and sex chromosomes.
7. People with red hair usually have very light skin. What might be a genetic explanation for this observation?

Multimedia resources

Points to Consider

You read in this lesson about the chromosomes and genes that control human traits. Most traits are controlled by genes on autosomes, but many are controlled by genes on the X chromosome.

- Do you think it matters whether a gene is on an autosome or the X chromosome when it comes to how it is inherited?
- How do mothers and fathers pass their sex chromosomes to their sons and daughters? Their autosomes?

8.2 Human Inheritance

Lesson Objectives

- Describe inheritance in humans for autosomal and X-linked traits.
- Identify complex modes of human inheritance.
- Describe genetic disorders caused by mutations or abnormal numbers of chromosomes.

Vocabulary

epistasis situation in which one gene affects the expression of another gene
gene therapy way to cure genetic disorders by inserting normal genes into cells with mutant genes

genetic trait characteristic that is encoded in DNA

multiple allele trait trait controlled by one gene with more than two alleles

nondisjunction failure of replicated chromosomes to separate during meiosis II, resulting in some gametes with a missing chromosome and some with an extra chromosome

pedigree chart showing how a trait is passed from generation to generation within a family

pleiotropy situation in which a single gene affects more than one trait

sex-linked trait traits controlled by a gene located on a sex chromosome

X-linked trait trait controlled by a gene located on the X chromosome

Introduction

Characteristics that are encoded in DNA are called **genetic traits**. Different types of human traits are inherited in different ways. Some human traits have simple inheritance patterns like the traits that Gregor Mendel studied in pea plants. Other human traits have more complex inheritance patterns.

Mendelian Inheritance in Humans

Mendelian inheritance refers to the inheritance of traits controlled by a single gene with two alleles, one of which may be dominant to the other. Not many human traits are controlled by a single gene with two alleles, but they are a good starting point for understanding human heredity. How Mendelian traits are inherited depends on whether the traits are controlled by genes on autosomes or the X chromosome.

Autosomal Traits

Autosomal traits are controlled by genes on one of the 22 human autosomes. Consider earlobe attachment. A single autosomal gene with two alleles determines whether you have attached earlobes or free-hanging earlobes. The allele for free-hanging earlobes (F) is dominant to the allele for attached earlobes (f). Other single-gene autosomal traits include widow's peak and hitchhiker's thumb. The dominant and recessive forms of these traits are shown in **Figure** 8.5. Which form of these traits do you have? What are your possible genotypes for the traits? The chart in **Figure** 8.5 is called a **pedigree**. It shows how the earlobe trait was passed from generation to generation within a family. Pedigrees are useful tools for studying inheritance patterns.

You can watch a video explaining how pedigrees are used and what they reveal at this link: http://www.youtube.com/watch?v=HbIHjsn5cHo.

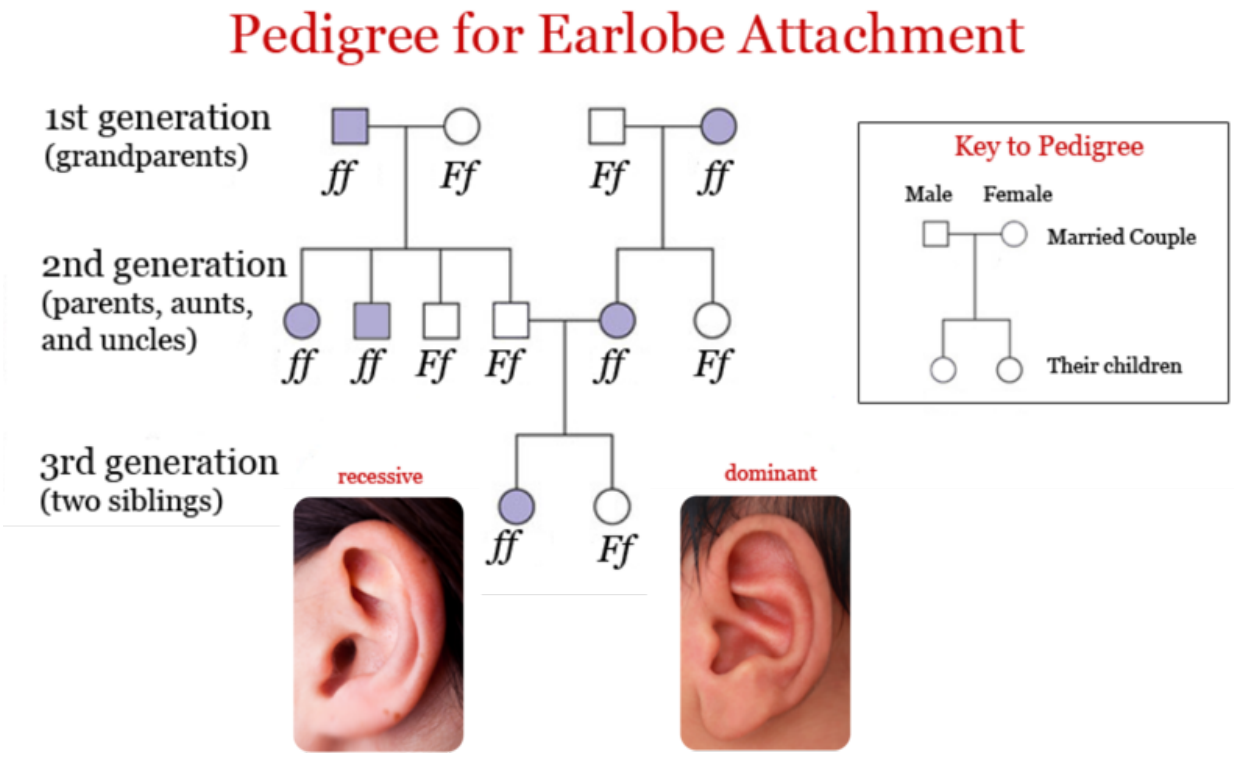


FIGURE 8.5

Having free-hanging earlobes is an autosomal dominant trait. This figure shows the trait and how it was inherited in a family over three generations. Shading indicates people who have the recessive form of the trait. Look at (or feel) your own earlobes. Which form of the trait do you have? Can you tell which genotype you have?

Other single-gene autosomal traits include widow's peak and hitchhiker's thumb. The dominant and recessive forms of these traits are shown in **Figure** 8.6. Which form of these traits do you have? What are your possible genotypes for the traits?

FIGURE 8.6

Widow's peak and hitchhiker's thumb are dominant traits controlled by a single autosomal gene.

Sex-Linked Traits

Traits controlled by genes on the sex chromosomes are called **sex-linked traits**, or **X-linked traits** in the case of the X chromosome. Single-gene X-linked traits have a different pattern of inheritance than single-gene autosomal traits. Do you know why? It's because males have just one X chromosome. In addition, they always inherit their X chromosome from their mother, and they pass it on to all their daughters but none of their sons. This is illustrated in **Figure** 8.7.

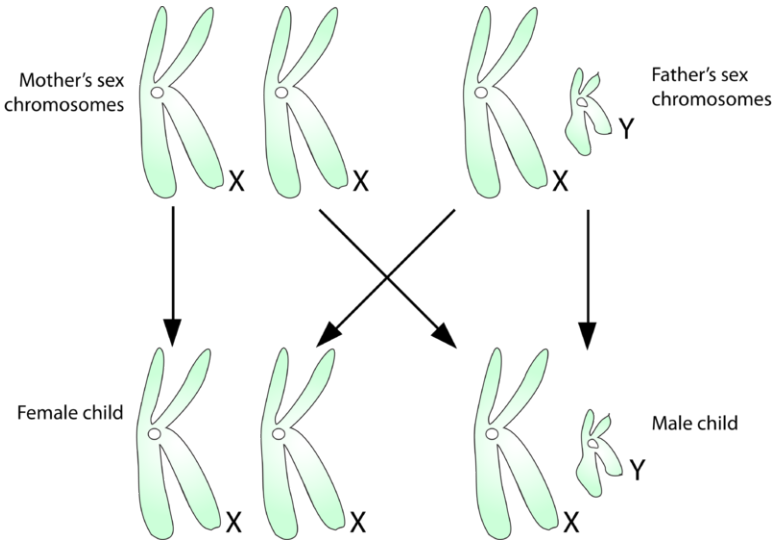


FIGURE 8.7

Inheritance of Sex Chromosomes. Mothers pass only X chromosomes to their children. Fathers always pass their X chromosome to their daughters and their Y chromosome to their sons. Can you explain why fathers always determine the sex of the offspring?

Because males have just one X chromosome, they have only one allele for any X-linked trait. Therefore, a recessive

X-linked allele is always expressed in males. Because females have two X chromosomes, they have two alleles for any X-linked trait. Therefore, they must inherit two copies of the recessive allele to express the recessive trait. This explains why X-linked recessive traits are less common in females than males. An example of a recessive X-linked trait is red-green color blindness. People with this trait cannot distinguish between the colors red and green. More than one recessive gene on the X chromosome codes for this trait, which is fairly common in males but relatively rare in females (**Figure** 8.8). At the link below, you can watch an animation about another X-linked recessive trait called hemophilia A. http://www.dnalc.org/view/16315-Animation-13-Mendelian-laws-apply-to-human-beings-.html

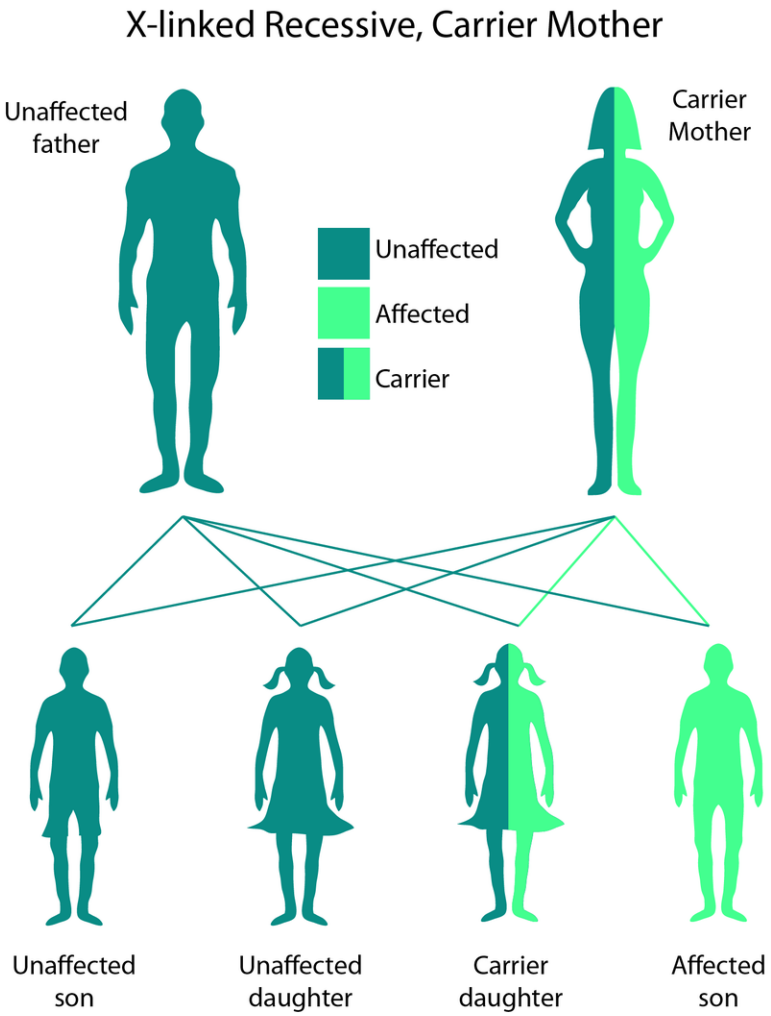
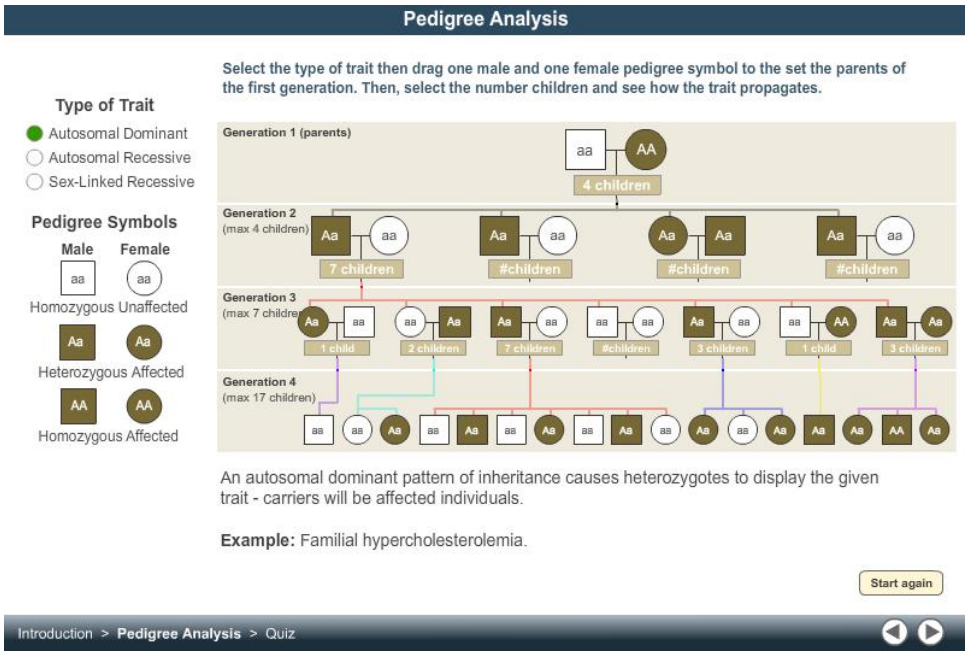


FIGURE 8.8

Pedigree for Color Blindness. Color blindness is an X-linked recessive trait. Mothers pass the recessive allele for the trait to their sons, who pass it to their daughters.

Pedigree Analysis Activity

The following link is to a pedigree analysis activity. Autosomal dominant, autosomal recessive and sex-linked recessive inheritance is explored through an interactive activity. CK-12 Pedigree Analysis Animation



Non-Mendelian Inheritance

Most human traits have more complex modes of inheritance than simple Mendelian inheritance. For example, the traits may be controlled by multiple alleles or multiple genes.

Multiple Allele Traits

The majority of human genes are thought to have more than two alleles. Traits controlled by a single gene with more than two alleles are called **multiple allele traits**. An example is ABO blood type. There are three common alleles for this trait, which can be represented by the letters A, B, and O. As shown in **Table** 8.1, there are six possible ABO genotypes but only four phenotypes. This is because alleles A and B are codominant to each other and both are dominant to O. You can learn more about ABO blood type by watching the video at this link: http://www.youtube.com/watch?v=oz4Ctau8mC8 (13:15).

TABLE 8.1: ABO Blood Type

Genotype	Phenotype
AA	A
AO	A
AB	AB
BB	B
BO	B
OO	O

Polygenic Traits

Many human traits are controlled by more than one gene. These traits are called polygenic traits (or characteristics). The alleles of each gene have a minor additive effect on the phenotype. There are many possible combinations of alleles, especially if each gene has multiple alleles. Therefore, a whole continuum of phenotypes is possible. An