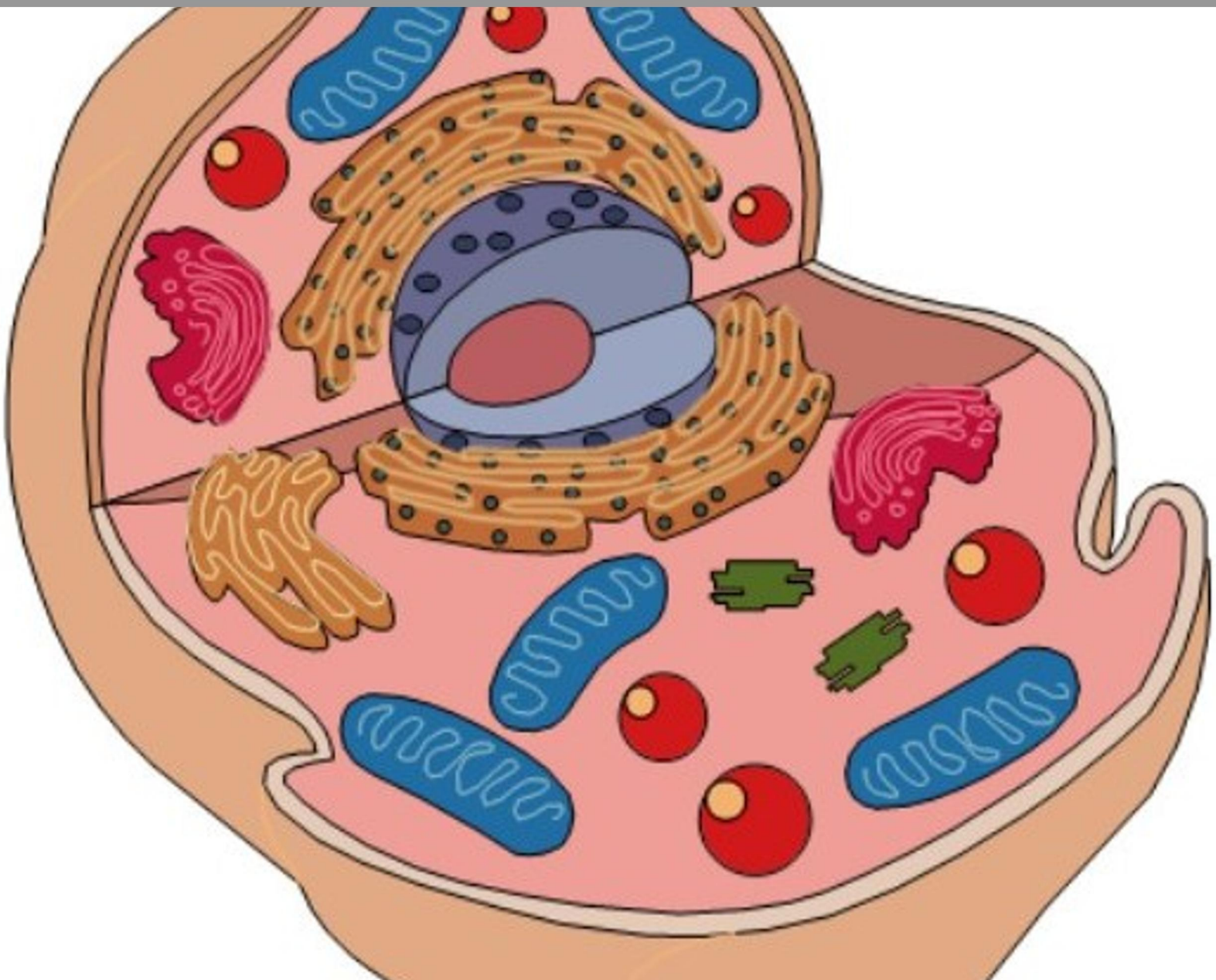


Science 9: Reproduction



Science 9: Cells & Reproduction

Peter MacDonald
Jessica Harwood
Douglas Wilkin, Ph.D.

Say Thanks to the Authors

Click <http://www.ck12.org/saythanks>

(No sign in required)

ck-12 **flexbook**
next generation textbooks

To access a customizable version of this book, as well as other interactive content, visit www.ck12.org

CK-12 Foundation is a non-profit organization with a mission to reduce the cost of textbook materials for the K-12 market both in the U.S. and worldwide. Using an open-source, collaborative, and web-based compilation model, CK-12 pioneers and promotes the creation and distribution of high-quality, adaptive online textbooks that can be mixed, modified and printed (i.e., the FlexBook® textbooks).

Copyright © 2016 CK-12 Foundation, www.ck12.org

The names “CK-12” and “CK12” and associated logos and the terms “**FlexBook®**” and “**FlexBook Platform®**” (collectively “CK-12 Marks”) are trademarks and service marks of CK-12 Foundation and are protected by federal, state, and international laws.

Any form of reproduction of this book in any format or medium, in whole or in sections must include the referral attribution link <http://www.ck12.org/saythanks> (placed in a visible location) in addition to the following terms.

Except as otherwise noted, all CK-12 Content (including CK-12 Curriculum Material) is made available to Users in accordance with the Creative Commons Attribution-Non-Commercial 3.0 Unported (CC BY-NC 3.0) License (<http://creativecommons.org/licenses/by-nc/3.0/>), as amended and updated by Creative Commons from time to time (the “CC License”), which is incorporated herein by this reference.

Complete terms can be found at <http://www.ck12.org/about/terms-of-use>.

Printed: October 3, 2016

flexbook
next generation textbooks



AUTHORS

Peter MacDonald
Jessica Harwood
Douglas Wilkin, Ph.D.

EDITOR

Douglas Wilkin, Ph.D.

CONTRIBUTORS

Doris Kraus, Ph.D.
Niamh Gray-Wilson
Jean Brainard, Ph.D.
Sarah Johnson
Jane Willan
Corliss Karasov

Contents

1	Characteristics of Life	1
2	Domains of Life	6
3	Cell Biology	10
4	Prokaryotic and Eukaryotic Cells	14
5	Cell Membrane	18
6	Cell Nucleus	21
7	Organelles	24
8	Cell Division	28
9	Cell Cycle	31
10	Meiosis	34
11	Mitosis vs. Meiosis	39
12	Asexual vs. Sexual Reproduction	42
13	Mendel’s Pea Plants	47
14	Mendel’s Laws and Genetics	51
15	Punnett Squares	54
16	DNA, the Genetic Material	57
17	DNA Structure and Replication	61
18	Mutations	64
19	Modern Genetics	68
20	Polygenic Traits	71
21	Sex-linked Inheritance	74
22	Genetic Disorders	78
23	Chromosomal Disorders	81

24 Cloning	85
25 Human Genome Project	88
26 Gene Therapy	91
27 Biotechnology in Agriculture	94

CONCEPT

1

Characteristics of Life

- Define what it means to be living.
- Know the five characteristics of living organisms.
- Describe the five characteristics shared by all living organisms.
- Identify the role of the five characteristics shared by all living organisms.
- Summarize in detail the role of each characteristic in life.



Is fire alive?

Fire can grow. Fire needs fuel and oxygen. But fire is not a form of life, although it shares a few traits with some living things. How can you distinguish between non-living and living things?

The Characteristics of Life

How do you define a living thing? What do mushrooms, daisies, cats, and bacteria have in common? All of these are living things, or **organisms**. It might seem hard to think of similarities among such different organisms, but they actually have many properties in common. Living organisms are similar to each other because all organisms evolved from the same common ancestor that lived billions of years ago.

All living organisms:

1. Need energy to carry out life processes.
2. Are composed of one or more cells.

3. Respond to their environment.
4. Grow and reproduce.
5. Maintain a stable internal environment.

Living Things Need Resources and Energy

Why do you eat everyday? To get energy. **Energy** is the ability to do work. Without energy, you could not do any "work." Though not doing any "work" may sound nice, the "work" fueled by energy includes everyday activities, such as walking, writing, and thinking. But you are not the only one who needs energy. In order to grow and reproduce and carry out the other process of life, all living organisms need energy. But where does this energy come from?

The source of energy differs for each type of living thing. In your body, the source of energy is the food you eat. Here is how animals, plants, and fungi obtain their energy:

- All animals must eat in order to obtain energy. Animals also eat to obtain building materials. Animals eat plants and other animals.
- Plants don't eat. Instead, they use energy from the sun to make their "food" through the process of **photosynthesis**.
- Mushrooms and other fungi obtain energy from other organisms. That's why you often see fungi growing on a fallen tree; the rotting tree is their source of energy (**Figure 1.1**).

Since plants harvest energy from the sun and other organisms get their energy from plants, nearly all the energy of living things initially comes from the sun.



FIGURE 1.1

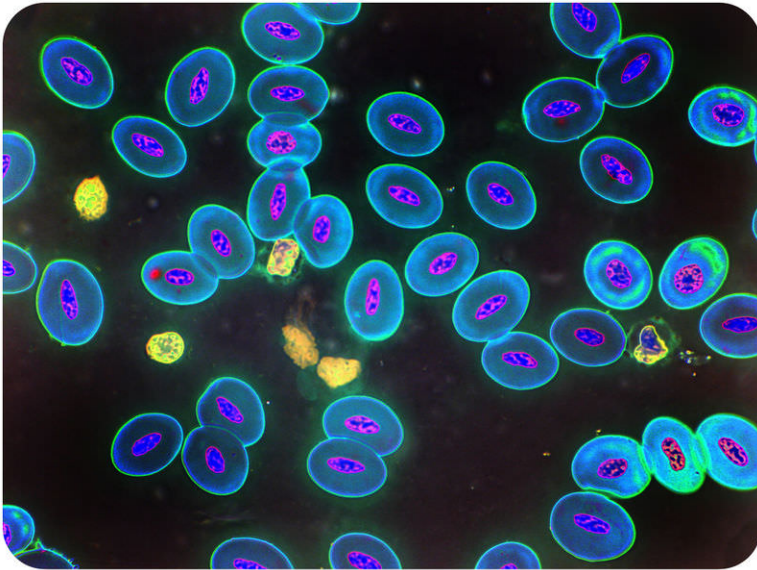
Orange bracket fungi on a rotting log in the Oak Openings Preserve in Ohio. Fungi obtain energy from breaking down dead organisms, such as this rotting log.

Living Things Are Made of Cells

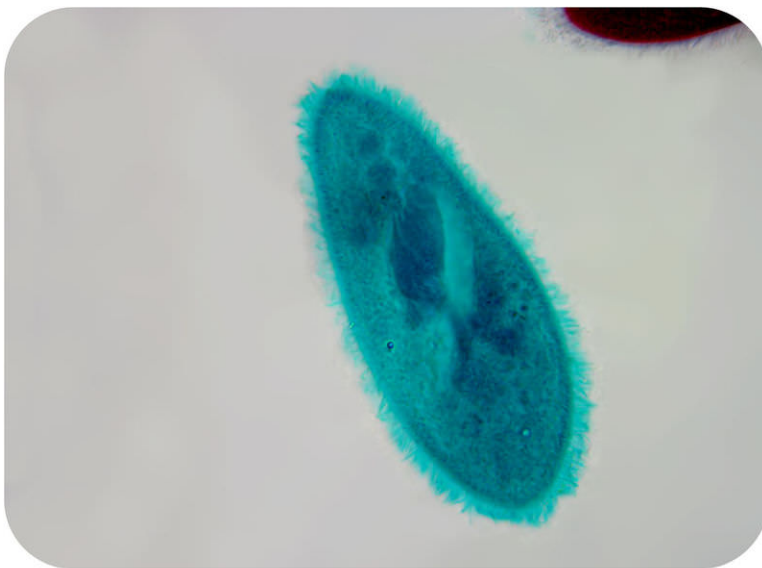
If you zoom in very close on a leaf of a plant, or on the skin on your hand, or a drop of blood, you will find cells, you will find cells (**Figure 1.2**). **Cells** are the smallest structural and functional unit of all living organisms. Most cells are so small that they are usually visible only through a microscope. Some organisms, like bacteria, plankton that live in the ocean, or the *Paramecium*, shown in **Figure 1.3**, are unicellular, made of just one cell. Other organisms have millions, billions, or trillions of cells.

All cells have at least some structures in common, such as **ribosomes**, which are the sites where proteins are made. All cells also have **DNA** and **proteins**. The **nucleus** is clearly visible in the blood cells (**Figure 1.2**). The nucleus can be described as the "information center," containing the instructions (DNA) for making all the proteins in a cell, as well as how much of each protein to make. The nucleus is also the main distinguishing feature between the two general categories of cell, with cells known as prokaryotic lacking a nucleus.

Although the cells of different organisms are built differently, they all have certain general functions. Every cell must get energy from food, be able to grow and divide, and respond to its environment. More about cell structure and function will be discussed in additional concepts.

**FIGURE 1.2**

These cells show the characteristic nucleus. A few smaller cells are also visible. This image has been magnified 1000 times its real size.

**FIGURE 1.3**

This *Paramecium* is a single-celled organism.

Living Organisms Respond to their Environment

All living organisms are able to react to something important or interesting in their external environment. For example, living organisms constantly respond to their environment. They respond to changes in light, heat, sound, and chemical and mechanical contact. Organisms have means for receiving information, such as eyes, ears, taste buds, or other structures.

Living Things Grow and Reproduce

All living things **reproduce** to make the next generation. Organisms that do not reproduce will go extinct. As a result, there are no species that do not reproduce (**Figure 1.4**). Some organisms reproduce asexually (**asexual**

reproduction), especially single-celled organisms, and make identical copies (or clones) of themselves. Other organisms reproduce sexually (**sexual reproduction**), combining genetic information from two parents to make genetically unique offspring.



FIGURE 1.4

Like all living things, cats reproduce to make a new generation of cats.

Living Things Maintain Stable Internal Conditions

When you are cold, what does your body do to keep warm? You shiver to warm up your body. When you are too warm, you sweat to release heat. When any living organism gets thrown off balance, its body or cells help it return to normal. In other words, living organisms have the ability to keep a stable internal environment. Maintaining a balance inside the body or cells of organisms is known as **homeostasis**. Like us, many animals have evolved behaviors that control their internal temperature. A lizard may stretch out on a sunny rock to increase its internal temperature, and a bird may fluff its feathers to stay warm (**Figure 1.5**).



FIGURE 1.5

A bird fluffs its feathers to stay warm and to maintain homeostasis.

Summary

- Living things are called organisms.
- All living organisms need energy to carry out life processes, are composed of one or more cells, respond to their environment, grow, reproduce, and maintain a stable internal environment.

Explore More

Use the resource below to answer the questions that follow.

- **Characteristics of Life** at http://www.youtube.com/watch?v=gJd65_Xrxs4 (3:15)
1. What are cell products? Do you think they should be included in characteristics of life? Why or why not?
 2. Are all responses to the environment immediately obvious? Be specific and explain your reasoning.
 3. Explain the concept of homeostasis. Give an example.
 4. At what level does life evolve?

Review

1. Is a crystal alive? Why or why not?
2. What is a cell?
3. What is homeostasis?
4. What are the two forms of reproduction? Describe the examples in your response.

References

1. Benny Mazur. [Fungi breaking down a rotting log](#) . CC BY 2.0
2. Image copyright Jubal Harshaw, 2014. [Nuclei of reptilian blood cells](#) . Used under license from Shutterstock.com
3. Image copyright Jubal Harshaw, 2014. [Picture of a paramecium, a single-celled organism](#) . Used under license from Shutterstock.com
4. Charles Nadeau. [Like all living things, cats reproduce to make a new generation of cats](#) . CC BY 2.0
5. Tony Hisgett. [A bird fluffs its feathers to stay warm and to maintain homeostasis](#) . CC BY 2.0

CONCEPT

2

Domains of Life

- Distinguish between the three domains of life.
- List the four Eukarya kingdoms.

**What do you have in common with pond scum?**

Humans are in the same domain as trees and algae, which makes up the "pond scum" you see here. What could they possibly have in common? It is the location of their DNA inside their cells. Their cells all have a nucleus that is home to their genetic material.

The Domains of Life

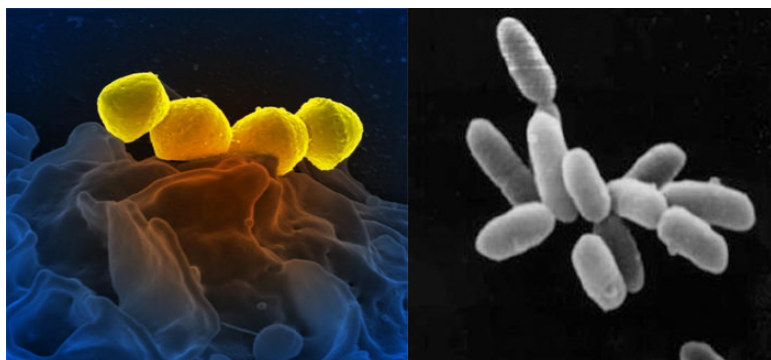
Let's explore the **domain**, the least specific category of classification.

All of life can be divided into three domains, based on the type of cell of the organism:

1. **Bacteria**: cells do not contain a nucleus.
2. **Archaea**: cells do not contain a nucleus; they have a different cell wall from bacteria.
3. **Eukarya**: cells do contain a nucleus.

Archaea and Bacteria

The Archaea and Bacteria domains (**Figure 2.1**) are both entirely composed of small, single-celled organisms and seem very similar, but they also have significant differences. Both are composed of **prokaryotic cells**, which are cells without a nucleus. In addition, both domains are composed of species that reproduce asexually (**asexual reproduction**) by dividing in two. Both domains also have species with cells surrounded by a **cell wall**, however, the cell walls are made of different materials. Bacterial cell walls contain the polysaccharide **peptidoglycan**. Lastly, Archaea often live in extreme environments including hot springs, geysers, and salt flats. Bacteria do not live in these environments.

**FIGURE 2.1**

The Group A *Streptococcus* organism (*left*) is in the domain Bacteria, one of the three domains of life. The *Halobacterium* (*right*) is in the domain Archaea, another one of the three domains.

Eukarya

All of the cells in the domain Eukarya keep their genetic material, or **DNA**, inside the **nucleus**. The domain Eukarya is made up of four kingdoms:

1. **Plantae**: Plants, such as trees and grasses, survive by capturing energy from the sun, a process called **photosynthesis**.
2. **Fungi**: Fungi, such as mushrooms and molds, survive by "eating" other organisms or the remains of other organisms. These organisms absorb their nutrients from other organisms.
3. **Animalia**: Animals also survive by eating other organisms or the remains of other organisms. Animals range from tiny ants to the largest whales, and include arthropods, fish, amphibians, reptiles, and mammals (**Figure 2.2**).
4. **Protista**: Protists are not all descended from a single common ancestor in the way that plants, animals, and fungi are. Protists are all the eukaryotic organisms that do not fit into one of the other three kingdoms. They include many kinds of microscopic one-celled (unicellular) organisms, such as algae and plankton, but also giant seaweeds that can grow to be 200 feet long.

Plants, animals, fungi, and protists might seem very different, but remember that if you look through a microscope, you will find similar cells with a membrane-bound nucleus in all of them. These are **eukaryotic cells**. These cells also have membrane-bound **organelles**, which prokaryotic cells lack. The main characteristics of the three domains of life are summarized in **Table 2.1**.

TABLE 2.1: Characteristics of the Three Domains of Life

	Archaea	Bacteria	Eukarya
Multicellular	No	No	Yes
Cell wall	Yes, without peptidoglycan	Yes, with peptidoglycan	Varies. Plants and fungi have a cell wall; animals do not.
Nucleus (Membrane-Enclosed DNA)	No	No	Yes
Membrane-Bound Organelles	No	No	Yes

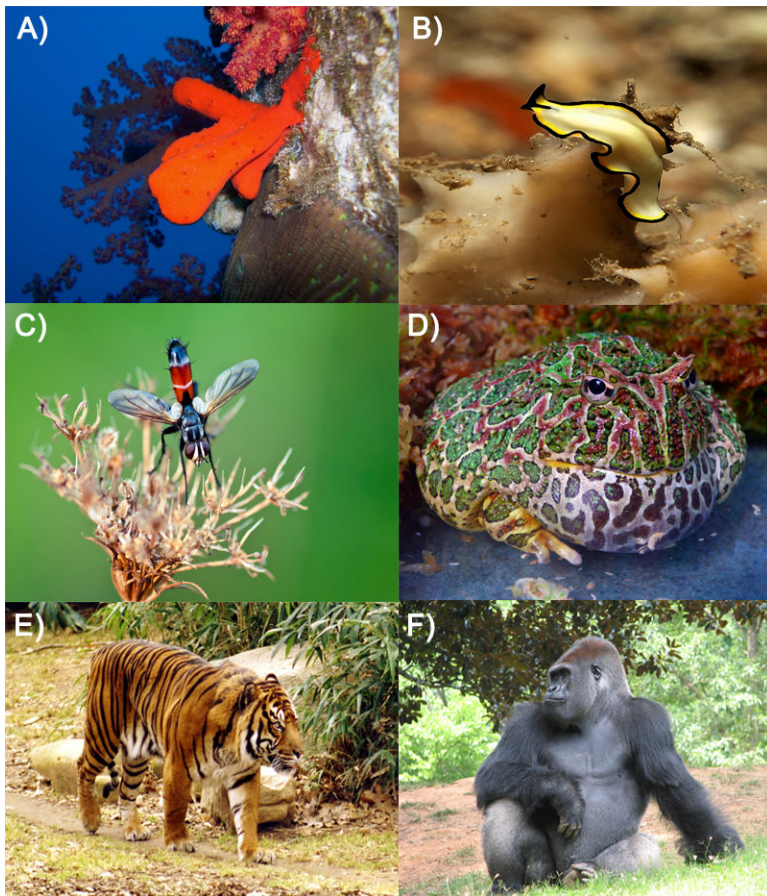


FIGURE 2.2

Diversity of Animals. These photos give just an inkling of the diversity of organisms that belong to the animal kingdom. (A) Sponge, (B) Flatworm, (C) Flying Insect, (D) Frog, (E) Tiger, (F) Gorilla.

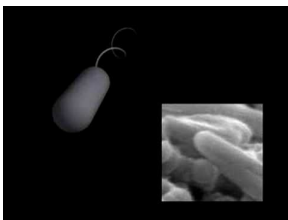
Summary

- All life can be classified into three domains: Bacteria, Archaea, and Eukarya.
- Organisms in the domain Eukarya keep their genetic material in a nucleus and include the plants, animals, fungi, and protists.

Explore More

Use the resource below to answer the questions that follow.

- **Exploring Deep-Subsurface: Life Domains** at <http://www.youtube.com/watch?v=UI7Yvu4McDU> (8:02)



MEDIA

Click image to the left or use the URL below.

URL: <https://www.ck12.org/flx/render/embeddedobject/57461>

1. What are the three domains of life?

2. What category do the individual organisms that we can see with our naked eye fall into?
3. What is an extremophile? What domain is known for these organisms? (Note: recent work has shown that extremophiles are not the only members of this domain.)
4. How do Archaea and Bacteria differ? How are they the same?
5. Which domain of life seems to be absent for deep-subsurface communities?

Review

1. Compare and contrast the domains Archaea and Bacteria.
2. What are the four kingdoms that make up the domain Eukarya?
3. Name three different examples of organisms in the domain Eukarya.

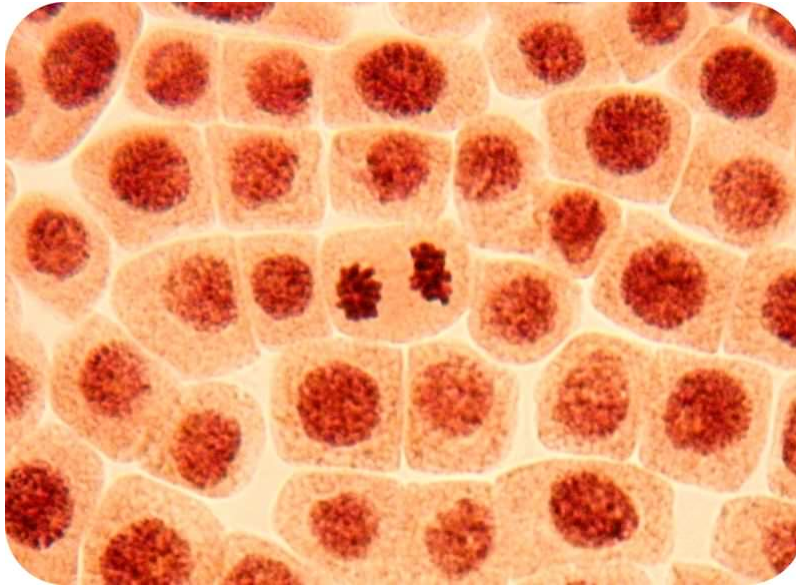
References

1. NIAID; NASA. [Pictures of bacteria and archaea](#) . CC BY 2.0; Public Domain
2. tsnoni; Stephen Childs; Umberto Salvagnin; Rusty Clark, Jeff Kubina, brokinhrt2. [Picture showing the diversity of animals](#) . CC BY 2.0

CONCEPT 3

Cell Biology

- Explain how cells are observed.
- Define cell. Describe the general role of a cell.
- State the three main parts of the cell theory.
- Summarize the structure-function relationship of a cell.
- Explain the levels of organization in an organism.



What are you made of?

Cells make up all living things, including your own body. This picture shows a typical group of cells. But not all cells look alike. Cells can differ in shape and sizes. And the different shapes usually means different functions.

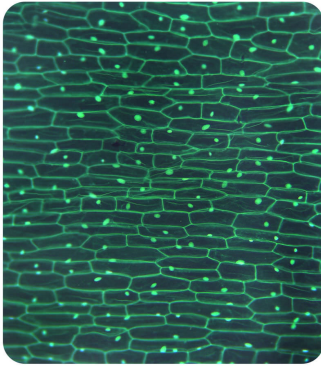
Introduction to Cells

A **cell** is the smallest structural and functional unit of an organism. Some organisms, like bacteria, consist of only one cell. Big organisms, like humans, consist of trillions of cells. Compare a human to a banana. On the outside, they look very different, but if you look close enough you'll see that their cells are actually very similar.

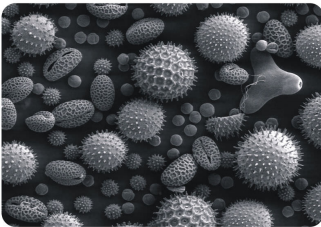
Observing Cells

Most cells are so small that you cannot see them without the help of a **microscope**. It was not until 1665 that English scientist Robert Hooke invented a basic light microscope and observed cells for the first time, by looking at a piece of cork. You may use light microscopes in the classroom. You can use a light microscope to see cells (**Figure 3.1**). But many structures in the cell are too small to see with a light microscope. So, what do you do if you want to see the tiny structures inside of cells?

In the 1950s, scientists developed more powerful microscopes. A light microscope sends a beam of light through a specimen, or the object you are studying. A more powerful microscope, called an **electron microscope**, passes a beam of electrons through the specimen. Sending electrons through a cell allows us to see its smallest parts, even the parts inside the cell (**Figure 3.2**). Without electron microscopes, we would not know what the inside of a cell looked like.

**FIGURE 3.1**

The outline of onion cells are visible under a light microscope.

**FIGURE 3.2**

An electron microscope allows scientists to see much more detail than a light microscope, as with this sample of pollen.

Cell Theory

In 1858, after using microscopes much better than Hooke's first microscope, Rudolf Virchow developed the hypothesis that cells only come from other cells. For example, bacteria, which are single-celled organisms, divide in half (after they grow some) to make new bacteria. In the same way, your body makes new cells by dividing the cells you already have. In all cases, cells only come from cells that have existed before. This idea led to the development of one of the most important theories in biology, the **cell theory**.

Cell theory states that:

1. All organisms are composed of cells.
2. Cells are alive and the basic living units of organization in all organisms.
3. All cells come from other cells.

As with other scientific theories, many hundreds, if not thousands, of experiments support the cell theory. Since Virchow created the theory, no evidence has ever been identified to contradict it.

Specialized Cells

Although cells share many of the same features and structures, they also can be very different (**Figure 3.3**). Each cell in your body is designed for a specific task. In other words, the cell's function is partly based on the cell's structure. For example:

- Red blood cells are shaped with a pocket that traps oxygen and brings it to other body cells.
- Nerve cells are long and stringy in order to form a line of communication with other nerve cells, like a wire. Because of this shape, they can quickly send signals, such as the feeling of touching a hot stove, to your brain.
- Skin cells are flat and fit tightly together to protect your body.

As you can see, cells are shaped in ways that help them do their jobs. Multicellular (many-celled) organisms have many types of specialized cells in their bodies.

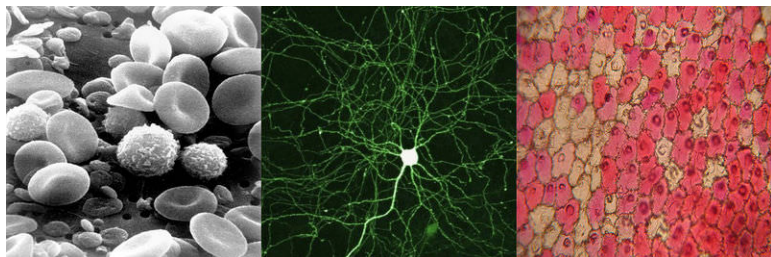


FIGURE 3.3

Red blood cells (*left*) are specialized to carry oxygen in the blood. Neurons (*center*) are shaped to conduct electrical impulses to many other nerve cells. These epidermal cells (*right*) make up the “skin” of plants. Note how the cells fit tightly together.

Levels of Organization

While cells are the basic units of an organism, groups of cells can perform a job together. These cells are called specialized because they have a special job. Specialized cells can be organized into **tissues**. For example, your liver cells are organized into liver tissue. Your liver tissue is further organized into an organ, your liver. **Organs** are formed from two or more specialized tissues working together to perform a job. All organs, from your heart to your liver, are made up of an organized group of tissues.

These organs are part of a larger system, the **organ systems**. For example, your brain works together with your spinal cord and other nerves to form the nervous system. This organ system must be organized with other organ systems, such as the circulatory system and the digestive system, for your body to work. Organ systems work together to form the entire organism. There are many levels of organization in living things (**Figure 3.4**).

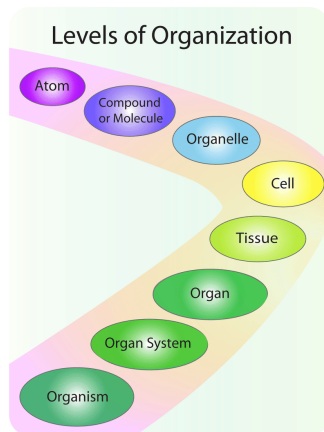


FIGURE 3.4

Levels of organization, from the atom (smallest) to the organism (largest). Notice that organelles are inside a cell, and organs are inside an organism.

Summary

- Cells were first observed under a light microscope, but today’s electron microscopes allow scientists to take a closer look at the inside of cells.
- Cell theory says that:
 - All organisms are composed of cells.

- Cells are alive and the basic living units of organization in all organisms.
- All cells come from other cells.
- Cells are organized into tissues, which are organized into organs, which are organized into organ systems, which are organized to create the whole organism.

Explore More

Use the sliding bar to zoom in on this animation to get an idea of the relative sizes of your cells.

- **Cell Size and Scale** at <http://learn.genetics.utah.edu/content/begin/cells/scale/>
1. What is the average size of a grain of salt?
 2. How big is an amoeba proteus? How big is a paramecium? (Remember this relationship for when you study amoeba.)
 3. How big is a skin cell? How big is a red blood cell? Can you think of any problems that might exist if this relationship was reversed? Explain your thinking fully.
 4. How big is an *E. coli* bacterium? How big is a mitochondrion? (Remember this relationship for when you study endosymbiosis.)
 5. Are all cells the same size?

Review

1. What type of microscope would be best for studying the structures found inside of cells?
2. What are the three basic parts of the cell theory?
3. According the cell theory, can you create a cell by combining molecules in a laboratory? Why or why not?
4. Give an example of a specialized cell.
5. What is a tissue?
6. What is the relationship between tissues and organs?

References

1. Image copyright Jubal Harshaw, 2014. [The outline of onion cells are visible under a light microscope](#) . Used under license from Shutterstock.com
2. Dartmouth Electron Microscope Facility. [An electron microscope image of pollen](#) . Public Domain
3. Bruce Wetzal and Harry Schaefer/National Cancer Institute; Mike Seyfang; Umberto Salvagnin. [Picture of red blood cells, neurons, and epidermal cells](#) . Public Domain; CC BY 2.0; CC BY 2.0
4. Rupali Raju. [Levels of organization in an organism](#) .

CONCEPT

4

Prokaryotic and Eukaryotic Cells

- Distinguish between eukaryotic and prokaryotic cells.
- Define an organelle.
- Describe the main role of the nucleus



Are bacteria cells like our cells?

Yes and no. Bacteria cells are similar to our cells in some ways. Like our cells, bacteria cells have DNA and a plasma membrane. But bacteria are unique in other ways. They are called prokaryotic cells because of these differences.

Prokaryotic and Eukaryotic

There are two basic types of cells, **prokaryotic cells** and **eukaryotic cells**. The main difference between eukaryotic and prokaryotic cells is that eukaryotic cells have a **nucleus**. The nucleus is where cells store their **DNA**, which is the genetic material. The nucleus is surrounded by a membrane. Prokaryotic cells do not have a nucleus. Instead, their DNA floats around inside the cell. Organisms with prokaryotic cells are called **prokaryotes**. All prokaryotes are single-celled (unicellular) organisms. Bacteria and Archaea are the only prokaryotes. Organisms with eukaryotic cells are called **eukaryotes**. Animals, plants, fungi, and protists are eukaryotes. All multicellular organisms are eukaryotes. Eukaryotes may also be single-celled.

Both prokaryotic and eukaryotic cells have structures in common. All cells have a plasma membrane, ribosomes, cytoplasm, and DNA. The **plasma membrane**, or cell membrane, is the phospholipid layer that surrounds the cell and protects it from the outside environment. **Ribosomes** are the non-membrane bound organelles where proteins are made, a process called **protein synthesis**. The **cytoplasm** is all the contents of the cell inside the cell membrane, not including the nucleus.

Eukaryotic Cells

Eukaryotic cells usually have multiple **chromosomes**, composed of DNA and protein. Some eukaryotic species have just a few chromosomes, others have close to 100 or more. These chromosomes are protected within the nucleus. In addition to a nucleus, eukaryotic cells include other membrane-bound structures called **organelles**. Organelles allow eukaryotic cells to be more specialized than prokaryotic cells. Pictured below are the organelles of eukaryotic cells (**Figure 7.1**), including the **mitochondria**, **endoplasmic reticulum**, and **Golgi apparatus**. These will be discussed in additional concepts.

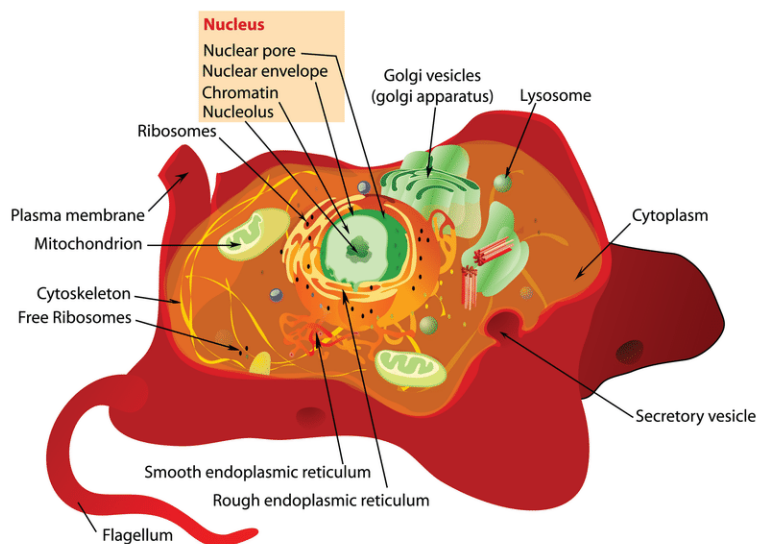


FIGURE 4.1

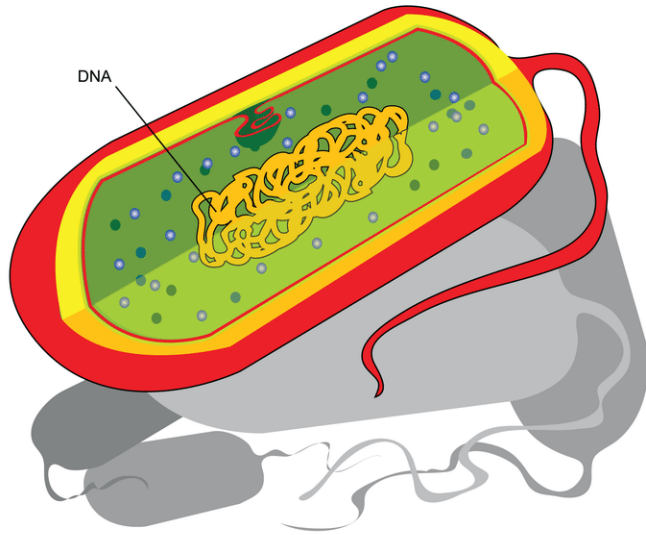
Eukaryotic cells contain a nucleus and various other special compartments surrounded by membranes, called organelles. The nucleus is where the DNA (chromatin) is stored. Organelles give eukaryotic cells more functions than prokaryotic cells.

Prokaryotic Cells

Prokaryotic cells (**Figure 4.2**) are usually smaller and simpler than eukaryotic cells. They do not have a nucleus or other membrane-bound organelles. In prokaryotic cells, the DNA, or genetic material, forms a single large circle that coils up on itself. The DNA is located in the main part of the cell.

TABLE 4.1: Comparison of Prokaryotic and Eukaryotic Cells

	Prokaryotic Cells	Eukaryotic Cells
Nucleus	No	Yes
DNA	Single circular piece of DNA	Multiple chromosomes
Membrane-Bound Organelles	No	Yes
Examples	Bacteria	Plants, animals, fungi


FIGURE 4.2

Prokaryotes do not have a nucleus. Instead, their genetic material is located in the main part of the cell.

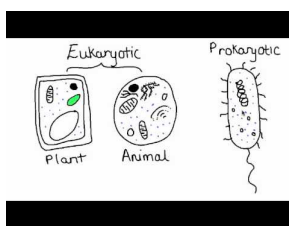
Summary

- All cells have a plasma membrane, ribosomes, cytoplasm, and DNA.
- Prokaryotic cells lack a nucleus and membrane-bound structures.
- Eukaryotic cells have a nucleus and membrane-bound structures called organelles.

Explore More

Use the resource below to answer the questions that follow.

- **Compare Prokaryotic and Eukaryotic Cells** at <http://www.youtube.com/watch?v=QON4z9vo7Ag> (1:55)



MEDIA

Click image to the left or use the URL below.

URL: <https://www.ck12.org/flx/render/embeddedobject/57353>

1. What does "naked" DNA mean? What kinds of organisms have "naked" DNA?
2. Where do you find membrane bound organelles? Are plasmids membrane bound organelles?
3. What is the function of mitochondria in prokaryotes?

Review

1. What do all cells have in common?
2. What are organelles?
3. Compare the location of the genetic material of eukaryotic cells and prokaryotic cells.

4. What are ribosomes?
5. What are the only prokaryotes?
6. Which prokaryotes are multicellular?

References

1. Mariana Ruiz Villarreal (LadyofHats), modified by CK-12 Foundation. [Organelles of a eukaryotic cell](#) .
2. Mariana Ruiz Villarreal (LadyofHats), modified by CK-12 Foundation. [Diagram of a prokaryotic cell](#) . Public Domain

CONCEPT **5**

Cell Membrane

- Describe the roles of the plasma membrane and cytosol.
- Explain the concept of semipermeability.
- Summarize how the plasma membrane separates the cytosol from the outside environment.



Who guards your cells?

Not everything can make it into your cells. Your cells have a plasma membrane that helps to guard your cells from unwanted intruders.

The Plasma Membrane and Cytosol

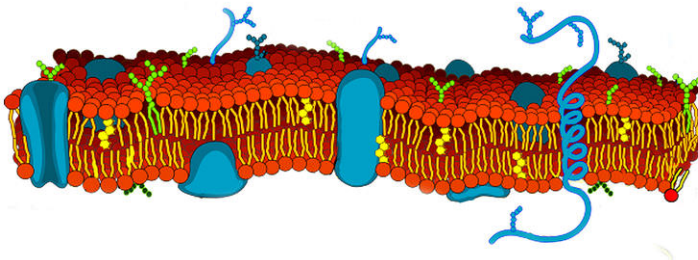
If the outside environment of a cell is water-based, and the inside of the cell is also mostly water, something has to make sure the cell stays intact in this environment. What would happen if a cell dissolved in water, like sugar does? Obviously, the cell could not survive in such an environment. So something must protect the cell and allow it to survive in its water-based environment. All cells have a barrier around them that separates them from the environment and from other cells. This barrier is called the **plasma membrane**, or cell membrane.

The Plasma Membrane

The plasma membrane (**Figure 5.1**) is made of a double layer of special lipids, known as **phospholipids**. The phospholipid is a lipid molecule with a hydrophilic ("water-loving") head and two hydrophobic ("water-hating") tails. Because of the hydrophilic and hydrophobic nature of the phospholipid, the molecule must be arranged in a specific pattern as only certain parts of the molecule can physically be in contact with water. Remember that there is water outside the cell, and the **cytoplasm** inside the cell is mostly water as well. So the phospholipids are arranged

in a double layer (a bilayer) to keep the cell separate from its environment. Lipids do not mix with water (recall that oil is a lipid), so the phospholipid bilayer of the cell membrane acts as a barrier, keeping water out of the cell, and keeping the cytoplasm inside the cell. The cell membrane allows the cell to stay structurally intact in its water-based environment.

The function of the plasma membrane is to control what goes in and out of the cell. Some molecules can go through the cell membrane to enter and leave the cell, but some cannot. The cell is therefore not completely permeable. "Permeable" means that anything can cross a barrier. An open door is completely permeable to anything that wants to enter or exit through the door. The plasma membrane is **semipermeable**, meaning that some things can enter the cell, and some things cannot.

**FIGURE 5.1**

Plasma membranes are primarily made up of phospholipids (orange). The hydrophilic ("water-loving") head and two hydrophobic ("water-hating") tails are shown. The phospholipids form a bilayer (two layers). The middle of the bilayer is an area without water. There can be water on either side of the bilayer. There are many proteins throughout the membrane.

Cytosol

The inside of all cells also contain a jelly-like substance called **cytosol**. Cytosol is composed of water and other molecules, including **enzymes**, which are proteins that speed up the cell's chemical reactions. Everything in the cell sits in the cytosol, like fruit in a jello mold. The term cytoplasm refers to the cytosol and all of the organelles, the specialized compartments of the cell. The cytoplasm does not include the nucleus. As a prokaryotic cell does not have a nucleus, the DNA is in the cytoplasm.

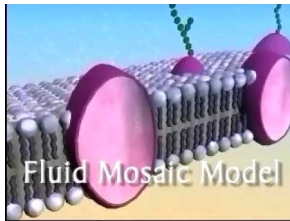
Summary

- The plasma membrane is formed by a phospholipid bilayer.
- The plasma membrane controls what moves inside and outside the cell.
- The cytosol is the jelly-like material in which the contents of the cell are suspended.

Explore More

Use the resource below to answer the following questions.

- **The Plasma Membrane** at <http://www.youtube.com/watch?v=moPJkCbKjBs> (5:16)



MEDIA

Click image to the left or use the URL below.

URL: <https://www.ck12.org/flx/render/embeddedobject/57347>

1. What makes up the "head" region of a phospholipid? Is it hydrophobic or hydrophilic?
2. What makes up the "tail" region of a phospholipid? Is it hydrophobic or hydrophilic?
3. What happens when you drop a phospholipid in water?
4. How are phospholipids arranged in a plasma membrane?
5. What is a glycoprotein? What is one of the uses of glycoproteins?
6. What is "Brownian movement"? How is this movement related to the cell membrane?

Review

1. What is the plasma membrane?
2. Describe a phospholipid.
3. What are the components of the cytosol?
4. What is meant by the description of the plasma membrane as "semipermeable"?
5. What is the difference between the cytosol and the cytoplasm?

References

1. Mariana Ruiz Villarreal (LadyofHats). [Drawing of a plasma membrane](#) . Public Domain

CONCEPT 6

Cell Nucleus

- Describe the features and function of the cell's nucleus.
- Define chromosome.
- Explain the role of the nucleolus.



Where is your DNA?

You may know that a criminal can easily leave DNA at a crime scene. How? DNA is found in every cell of your body. In each cell there is a nucleus, which is home to your DNA. So if a criminal has a cut, and blood is left at the crime scene, or a hair falls out and is left behind, then DNA will also be left at the scene.

The Nucleus

The **nucleus** is only found in eukaryotic cells. It contains most of the genetic material (the **DNA**) of the cell. The genetic material of the nucleus is like a set of instructions. These instructions tell the cell how to build molecules needed for the cell to function properly. That is, the DNA tells the cell how to build molecules needed for life. The nucleus is surrounded by the **nuclear envelope**, a double membrane (two phospholipid bilayers) that controls what goes in and out of the nucleus. The nucleus also has holes embedded in the nuclear envelope. These holes are known as **nuclear pores**, and they allow things to flow in and out of the nucleus.

Chromosomes

Inside of the nucleus, you will find the **chromosomes**. Chromosomes are strands of DNA wrapped around proteins. They contain **genes**, or small units of genetic material (DNA) that contains the code for the creation of a protein. Human cells have 46 chromosomes (23 pairs). There are hundreds to thousands of genes on each chromosome.

Nucleolus

The nucleus of many cells also contains a central region called the **nucleolus**. The job of the nucleolus is to build ribosomes. These ribosomes flow out the nuclear pores into the cytoplasm. **Ribosomes** are organelles that make proteins in the cytoplasm. See the composition of the nucleus pictured below (**Figure 6.1**).

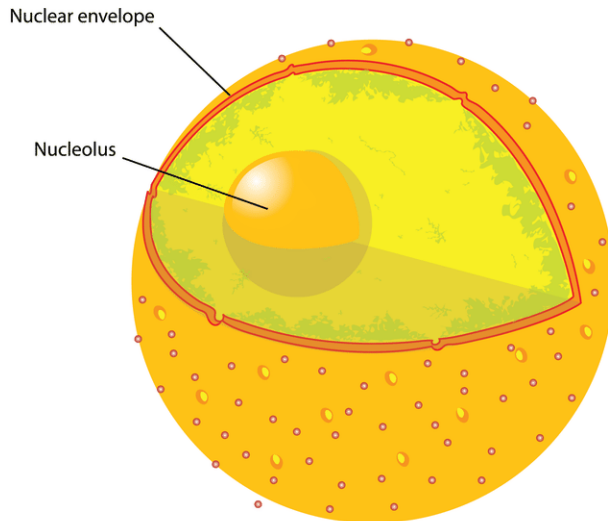


FIGURE 6.1

In eukaryotic cells, the DNA is kept in the nucleus. The nucleus is surrounded by a double membrane called the nuclear envelope. Within the nucleus is the nucleolus.

Summary

- The nucleus contains the genetic material of the cell.
- The genetic material of the cell is found in chromosomes, DNA wrapped around proteins.
- The nucleolus, which makes ribosomes, is also within the nucleus.

Explore More

Use the resources below to answer the following questions.

- **Nucleus** at <http://www.youtube.com/watch?v=DMd3mr6rQ2o> (2:25)
1. What is chromatin? What molecules make up chromatin?
 2. What is the nuclear membrane? What kind of membrane is this membrane?
 3. What is the nucleolus? What molecules do you find there?
 4. How is the inside of the nucleus connected to the cytosol? Why is this connection vital for the cell?

Review

1. What is contained in the nucleus of a cell?
2. What is a chromosome? Where are chromosomes located?
3. How many chromosomes do humans have?
4. What is the function of the nucleolus?

References

1. Mariana Ruiz Villarreal (LadyofHats), modified by CK-12 Foundation. [Position of the nucleus inside a cell](#) .
Public Domain

CONCEPT 7

Organelles

- List the main organelles found in an eukaryotic cell.
- Define the role of a ribosome.
- Describe the functions of the mitochondria, endoplasmic reticulum, and Golgi apparatus.
- Explain the function of a vesicle, a vacuole, and a lysosome.



Do brain cells have the same internal structures as your other cells?

Yes. Although brain cells look quite different from your other cells, they have the same internal structures as other cells. They need the same structures because they need to perform the same tasks, such as making proteins and obtaining energy.

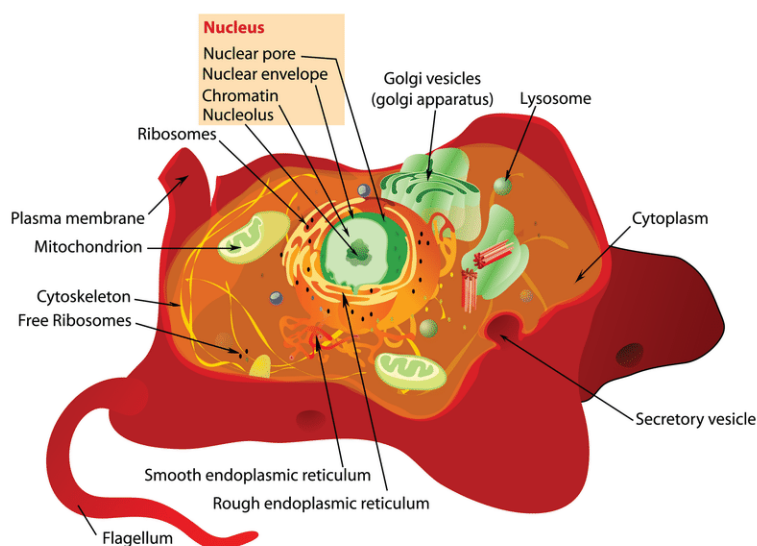
Organelles

Eukaryotic cells have many specific functions, so it can be said that a cell is like a factory. A factory has many machines and people, and each has a specific role. Just like a factory, the cell is made up of many different parts. Each part has a special role. The different parts of the cell are called **organelles**, which means "small organs." All organelles are found in eukaryotic cells. Prokaryotic cells are "simpler" than eukaryotic cells. Though prokaryotic cells still have many functions, they are not as specialized as eukaryotic cells, lacking membrane-bound organelles. Thus, most organelles are not found in prokaryotic cells.

Below are the main organelles found in eukaryotic cells (**Figure 7.1**):

1. The **nucleus** of a cell is like a safe containing the factory's trade secrets. The nucleus contains the genetic material (DNA), the information needed to build thousands of proteins.

- The **mitochondria** are the powerhouses of the cell. Mitochondria are the organelles where cellular energy is produced, providing the energy needed to power chemical reactions. This process, known as **cellular respiration**, produces energy in the form of **ATP** (adenosine triphosphate). Cells that use a lot of energy may have thousands of mitochondria.
- Vesicles** are small membrane bound sacs that transport materials around the cell and to the cell membrane.
- The **vacuoles** are like storage centers. Plant cells have larger vacuoles than animal cells. Plants store water and nutrients in their large central vacuoles.
- Lysosomes** are like the recycling trucks that carry waste away from the factory. Lysosomes have digestive enzymes that break down old molecules into parts that can be recycled.
- In both eukaryotes and prokaryotes, **ribosomes** are the non-membrane bound organelles where proteins are made. Ribosomes are like the machines in the factory that produce the factory's main product. Proteins are the main product of the cell.
- Some ribosomes can be found on folded membranes called the **endoplasmic reticulum** (ER), others float freely in the cytoplasm. If the ER is covered with ribosomes, it looks bumpy like sandpaper, and is called the rough endoplasmic reticulum. If the ER does not contain ribosomes, it is smooth and called the smooth endoplasmic reticulum. Many proteins are made on the ribosomes on the rough ER. These proteins immediately enter the ER, where they are modified, packaged into vesicles and sent to the Golgi apparatus. Lipids are made in the smooth ER.
- The **Golgi apparatus** works like a mail room. The Golgi apparatus receives proteins from the rough ER and puts "shipping addresses" on them. The Golgi then packages the proteins into vesicles and sends them to the right place in the cell or to the cell membrane. Some of these proteins are secreted from the cell (they exit the cell); others are placed into the cell membrane.


FIGURE 7.1

Eukaryotic cells contain special compartments surrounded by membranes, called organelles. For example, notice in this image the mitochondria, lysosomes, and Golgi apparatus.

Also, the **cytoskeleton** gives the cell its shape, and the **flagella** helps the cell to move. Prokaryotic cells may also have flagella.

Summary

- The nucleus stores the genetic information.
- The vacuoles are needed for storage.
- The lysosomes recycle waste.

- The cytoskeleton provides the shape of the cell.
- The ribosomes produce proteins.
- The rough ER is covered with ribosomes and makes proteins, while the smooth ER makes lipids.
- The Golgi apparatus packages proteins.

Explore More

Use the resources below to answer the following questions.

Explore More I

- **Cell Organelles and Their Function** at <http://www.youtube.com/watch?v=fKEaTt9heNM> (6:25)



MEDIA

Click image to the left or use the URL below.

URL: <https://www.ck12.org/flx/render/embeddedobject/114302>

1. What are the functions of the endoplasmic reticulum? What gives the rough endoplasmic reticulum its "rough" appearance?
2. What are the most abundant organelles in a cell? Where do they occur? What is their function?
3. What is the appearance of the Golgi apparatus? What is the function of the Golgi apparatus?
4. What are lysosomes? What are their functions?
5. What is the function of mitochondria? Do all cells have the same number of mitochondria? How can this situation be explained?

Explore More II

- **Plant and Animal Cell Organelles** at http://www.cellsalive.com/cells/cell_model.htm .

1. What is cytosol? How does this differ from cytoplasm?
2. What are the primary types of protein filaments that make up the cytoskeleton?
3. What is the function of a peroxisome?
4. What is a secretory vesicle? Where are they made? What is their function?

Review

1. What is the purpose of the Golgi apparatus?
2. What is the purpose of the mitochondria?
3. How is the smooth ER different from the rough ER?
4. What is a lysosome?

References

1. Mariana Ruiz Villarreal (LadyofHats), modified by CK-12 Foundation. [Organelles of a eukaryotic cell](#) .
Public Domain

CONCEPT 8

Cell Division

- Define cell division.
- Explain why cells must divide.



Do cells get worn out?

Yes, just like this car, cells cannot last forever. Cells do eventually wear out. At that point, they need to be replaced. This is one reason that your cells divide. New cells that result after cells divide are also used for growth and to repair cuts.

Why Cells Divide

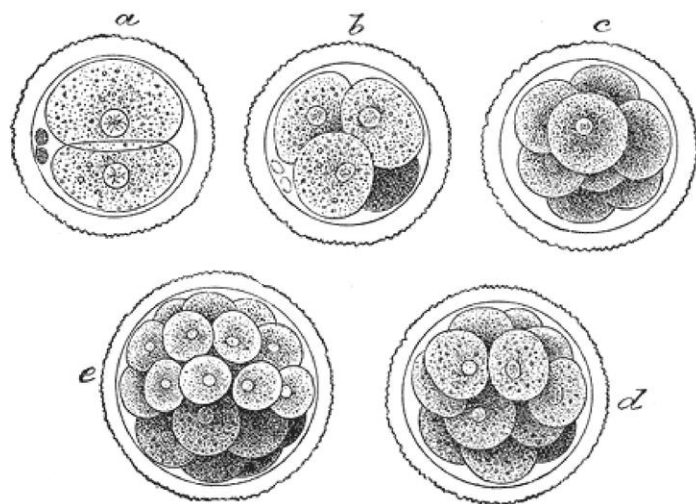
Imagine the first stages of life. In humans and other animals, a sperm fertilizes an egg, forming the first cell. But humans are made up of trillions of cells, so where do the new cells come from? Remember that according to the **cell theory**, all cells come from existing cells. Once a sperm and egg cell unite and the first cell, called a **zygote**, forms, an entire baby will develop. And each cell in that baby will be genetically identical, meaning that each cell will have exactly the same DNA.

How does a new life go from one cell to so many? The cell divides in half, creating two cells. Then those two cells divide, for a total of four cells. The new cells continue to divide and divide. One cell becomes two, then four, then eight, and so on (**Figure 8.1**). This continual process of a cell dividing and creating two new cells is known as **cell division**. Cell division is part of a cycle of cellular growth and division known as the cell cycle—cells must grow before they divide. The **cell cycle** describes the "life" of a eukaryotic cell. In addition to cell division, the cell cycle includes the division of the nucleus and the cytoplasm.

Most cell division produces genetically identical cells, meaning they have the same DNA. The process of **mitosis**, which specifically is the division of the nucleus, ensures that each cell has the same DNA. During mitosis, the chromosomes equally separate, thus making sure each nucleus in each resulting cell after cell division is genetically identical.

A special form of cell division, called **meiosis**, produces cells with half as much DNA as the parent cell. These cells are used for reproduction. In prokaryotic organisms, cell division is how those organisms reproduce.

Besides the development of a baby, there are many other reasons that cell division is necessary for life:

**FIGURE 8.1**

Cells divide repeatedly to produce an embryo. Previously the one-celled zygote (the first cell of a new organism) divided to make two cells (a). Each of the two cells divides to yield four cells (b), then the four cells divide to make eight cells (c), and so on. Through cell division, an entire embryo forms from one initial cell.

1. To grow and develop, you must form new cells. Imagine how often your cells must divide during a growth spurt. Growing just an inch requires countless cell divisions. Your body must produce new bone cells, new skin cells, new cells in your blood vessels and so on.
2. Cell division is also necessary to repair damaged cells. Imagine you cut your finger. After the scab forms, it will eventually disappear and new skin cells will grow to repair the wound. Where do these cells come from? Some of your existing skin cells divide and produce new cells.
3. Your cells can also simply wear out. Over time you must replace old and worn-out cells. Cell division is essential to this process.

Summary

- Cells must divide repeatedly for an embryo to develop or for you to grow.
- Cells also divide in order to replace damaged or worn-out cells.

Explore More

Use the resources below to answer the questions that follow.

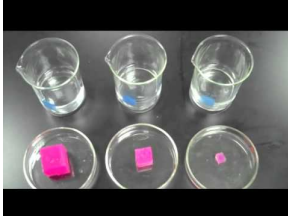
Explore More I

- **Why Must A Cell Divide** at <http://plaza.ufl.edu/alallen/pgl/modules/rio/stingarees/module/why.html>

1. What limits the size a cell can become? Be as specific as you can.
2. If you double the size of a cube, how does this affect the surface to volume ratio?

Explore More II

- **Surface Area to Volume Ratios** at <http://www.youtube.com/watch?v=xuG4ZZ1GbZI> (2:45)



MEDIA

Click image to the left or use the URL below.

URL: <https://www.ck12.org/flx/render/embeddedobject/57343>

1. Does the rate at which materials diffuse into a cell vary with the size of the cell?
2. What does this mean for large cells?

Review

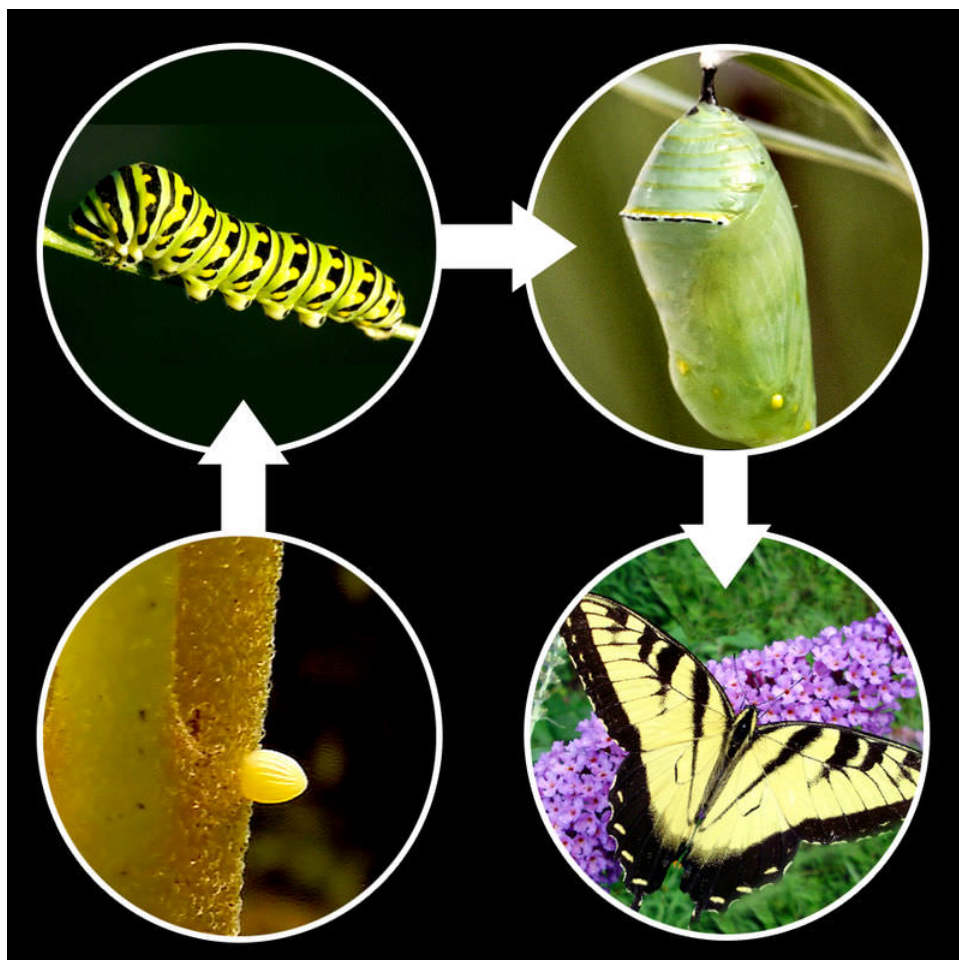
1. Define cell division.
2. How does an embryo develop from a fertilized egg?
3. List two reasons that cells must divide.

References

1. Gray's Anatomy of the Human Body by Henry Gray (1918). [Drawing of mitosis in a zygote to form an embryo](#) . Public Domain

CONCEPT **9****Cell Cycle**

- Describe the cell cycle.
- Explain the phases of the cell cycle.

**Do cells have a life cycle?**

Yes, just like a butterfly passes through different phases, such as caterpillar, chrysalis, and adult butterfly, there are a series of phases in a cell's life as it gets ready to divide. The sequence of phases leading up to cell division and then ending with cell division itself is called the cell cycle.

Cell Cycle

The process of cell division in eukaryotic cells is carefully controlled. The **cell cycle (Figure 9.1)** is the life cycle of an eukaryotic cell, with cell division at the end of the cycle. Like a human life cycle, which is made up of different phases, like childhood, adolescence, and adulthood, the cell cycle also occurs in a series of phases. The first cell cycle begins with the formation of a **zygote** from the fusion of a male and female sex cell (**gamete**).

The steps of the cell cycle can be divided into two main components: interphase and the mitotic phase. **Interphase** is the stage when the cell mostly performs its “everyday” functions. For example, it is when a kidney cell does what

a kidney cell is supposed to do. The cell also gets ready to divide during this time. The cell divides during the mitotic phase, which consists of mitosis and cytokinesis.

Most of the cell cycle consists of interphase, the time between cell divisions. Interphase can be divided into three stages:

1. The first growth phase (G1): During the G1 stage, the cell doubles in size and doubles the number of organelles.
2. The synthesis phase (S): The DNA is replicated during this phase. In other words, an identical copy of all the cell's DNA is made. This ensures that each new cell has a set of genetic material identical to that of the parental cell. This process is called **DNA replication**.
3. The second growth phase (G2): Proteins are synthesized that will help the cell divide. At the end of interphase, the cell is ready to enter mitosis.

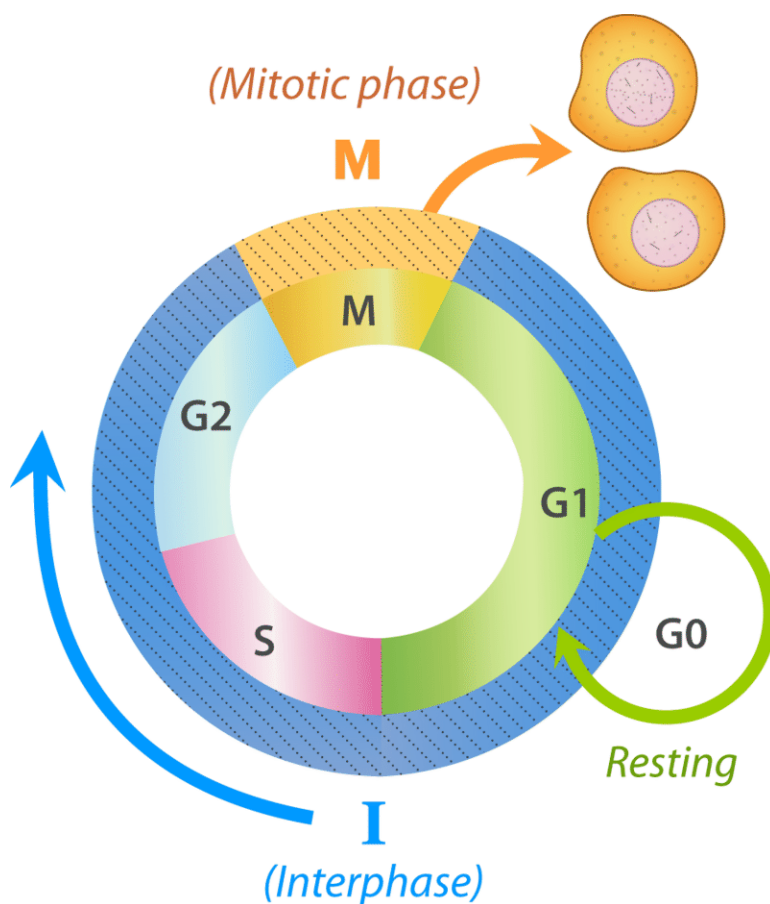


FIGURE 9.1

Shown is the cell cycle. Notice that most of the cell cycle is spent in Interphase (G1, S, and G2). Mitosis and cytokinesis occur during the Mitotic phase. Some cells may enter a resting phase during which progression through the cycle stops.

During **mitosis**, the nucleus divides as the chromosomes are equally separated. One nucleus becomes two nuclei, each with an identical set of **chromosomes**. Mitosis is followed by **cytokinesis**, when the cytoplasm divides,

resulting in two cells. After cytokinesis, cell division is complete. The one parent cell (the dividing cell) forms two genetically identical daughter cells (the cells that divide from the parent cell). The term "genetically identical" means that each cell has an identical set of DNA, and this DNA is also identical to that of the parent cell. If the cell cycle is not carefully controlled, it can cause a disease called **cancer** in which the cells divide out of control. A tumor can result from this kind of growth.

Summary

- The cell cycle describes the "life" of a cell.
- Interphase, the stage of the cell cycle when the cell, preparing to divide, is divided into the G1, S, and G2 stages.
- The nucleus divides during mitosis, and the cytoplasm divides during cytokinesis.

Explore More

Use the resource below to answer the questions that follow.

- **Cell Division and Cell Cycle** at <http://vimeo.com/9536315> (5:34)



MEDIA

Click image to the left or use the URL below.

URL: <https://www.ck12.org/flx/render/embeddedobject/57360>

1. What are the major phases of the cell cycle?
2. Why do you think it is important for a cell to grow before it replicates its DNA? Be as specific as you can in your answer.
3. What happens during the S phase of the cell cycle?
4. What happens during mitosis?
5. What is the function of spindle fibers? Where do they attach?
6. What is the function of meiosis?

Review

1. What is the cell cycle?
2. What are the two main components of the cell cycle?
3. What occurs during interphase?
4. What are the three stages of interphase?
5. What is the main purpose of the S phase?
6. Describe the main events of the mitotic phase.
7. Define cancer.

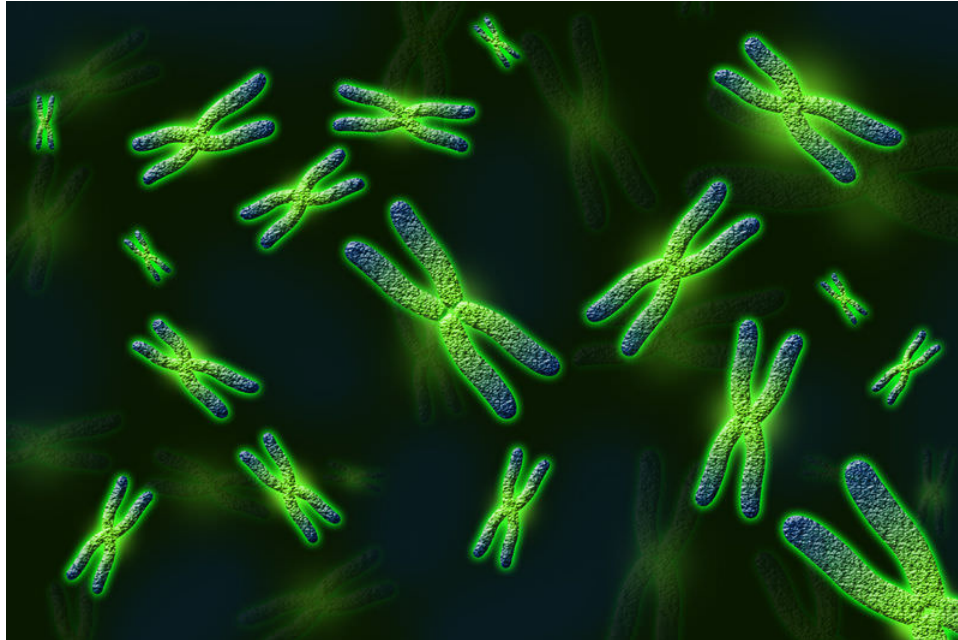
References

1. Hana Zavadska. [Diagram of the cell cycle](#) .

CONCEPT 10

Meiosis

- Explain the importance of meiosis.
- Distinguish between haploid and diploid.
- List the stages of meiosis.
- Summarize the steps of meiosis.
- Define crossing-over and explain its significance.



Do you have ALL your parents' chromosomes?

No, you only received half of your mother's chromosomes and half of your father's chromosomes. If you inherited them all, you would have twice the number of chromosomes that you're supposed to have. Humans typically have 23 pairs of chromosomes. If you received all your parents' chromosomes, you would have 46 pairs!

Introduction to Meiosis

Sexual reproduction combines gametes from two parents. **Gametes** are reproductive cells, such as sperm and egg. As gametes are produced, the number of chromosomes must be reduced by half. Why? The **zygote** must contain genetic information from the mother and from the father, so the gametes must contain half of the chromosomes found in normal body cells. When two gametes come together at fertilization, the normal amount of chromosomes results. Gametes are produced by a special type of cell division known as **meiosis**. Meiosis contains two rounds of cell division without DNA replication in between. This process reduces the number of chromosomes by half.

Human cells have 23 pairs of chromosomes, and each chromosome within a pair is called a **homologous chromosome**. For each of the 23 chromosome pairs, you received one chromosome from your father and one chromosome from your mother. **Alleles** are alternate forms of genes found on chromosomes. Homologous chromosomes have the same genes, though they may have different alleles. So, though homologous chromosomes are very similar, they are not identical. The homologous chromosomes are separated when gametes are formed. Therefore, gametes have only 23 chromosomes, not 23 pairs.

Haploid vs. Diploid

A cell with two sets of chromosomes is **diploid**, referred to as $2n$, where n is the number of sets of chromosomes. Most of the cells in a human body are diploid. A cell with one set of chromosomes, such as a gamete, is **haploid**, referred to as n . Sex cells are haploid. When a haploid sperm (n) and a haploid egg (n) combine, a diploid zygote will be formed ($2n$). In short, when a diploid zygote is formed, half of the DNA comes from each parent.

Overview of Meiosis

Before meiosis begins, DNA replication occurs, so each chromosome contains two sister chromatids that are identical to the original chromosome. Meiosis (**Figure 10.1**) is divided into two divisions: Meiosis I and Meiosis II. Each division can be divided into the same phases: prophase, metaphase, anaphase, and telophase. Cytokinesis follows telophase each time. Between the two cell divisions, DNA replication does not occur. Through this process, one diploid cell will divide into four haploid cells.

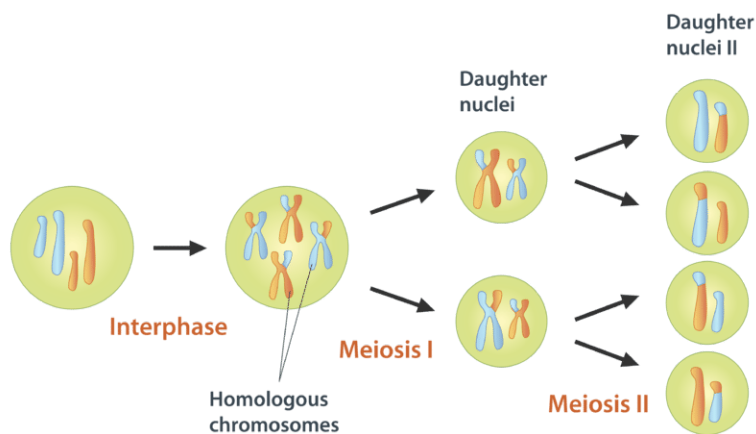


FIGURE 10.1

Overview of Meiosis. During meiosis, four haploid cells are created from one diploid parent cell.

Meiosis I

During meiosis I, the pairs of homologous chromosomes are separated from each other. This requires that they line up in their homologous pairs during metaphase I. The steps are outlined below:

1. Prophase I: The homologous chromosomes line up together. During this time, a process that only happens in meiosis can occur. This process is called **crossing-over** (**Figure 10.2**), which is the exchange of DNA between homologous chromosomes. Crossing-over forms new combinations of alleles on the resulting chromosome. Without crossing-over, the offspring would always inherit all of the alleles on one of the homologous chromosomes. Also during prophase I, the **spindle** forms, the chromosomes condense as they coil up tightly, and the nuclear envelope disappears.
2. Metaphase I: The homologous chromosomes line up in their pairs in the middle of the cell. Chromosomes from the mother or from the father can each attach to either side of the spindle. Their attachment is random, so all of the chromosomes from the mother or father do not end up in the same gamete. The gamete will contain some chromosomes from the mother and some chromosomes from the father.
3. Anaphase I: The homologous chromosomes are separated as the spindle shortens, and begin to move to opposite sides (opposite poles) of the cell.
4. Telophase I: The spindle fibers dissolve, but a new nuclear envelope does not need to form. This is because, after cytokinesis, the nucleus will immediately begin to divide again. No DNA replication occurs between

meiosis I and meiosis II because the chromosomes are already duplicated. After cytokinesis, two haploid cells result, each with chromosomes made of sister chromatids.

Since the separation of chromosomes into gametes is random during meiosis I, this process results in different combinations of chromosomes (and alleles) in each gamete. With 23 pairs of chromosomes, there is a possibility of over 8 million different combinations of chromosomes (2^{23}) in a human gamete.

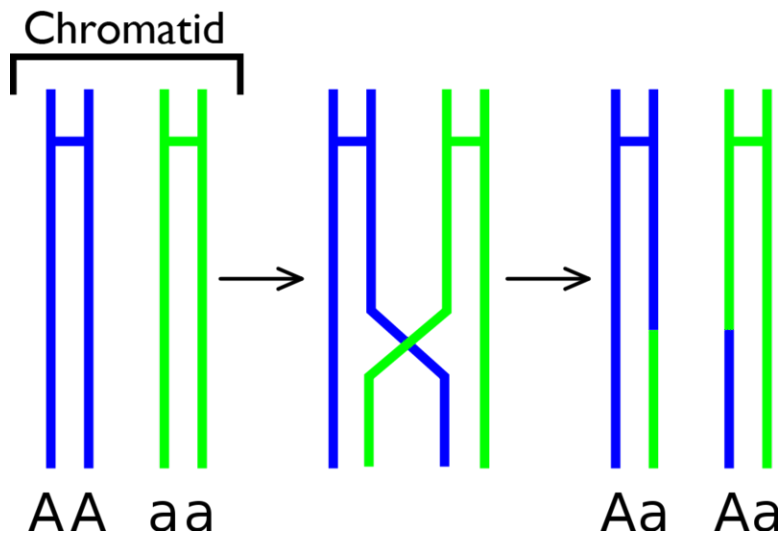


FIGURE 10.2

During crossing-over, segments of DNA are exchanged between non-sister chromatids of homologous chromosomes. Notice how this can result in an allele (A) on one chromatid being moved onto the other non-sister chromatid.

Meiosis II

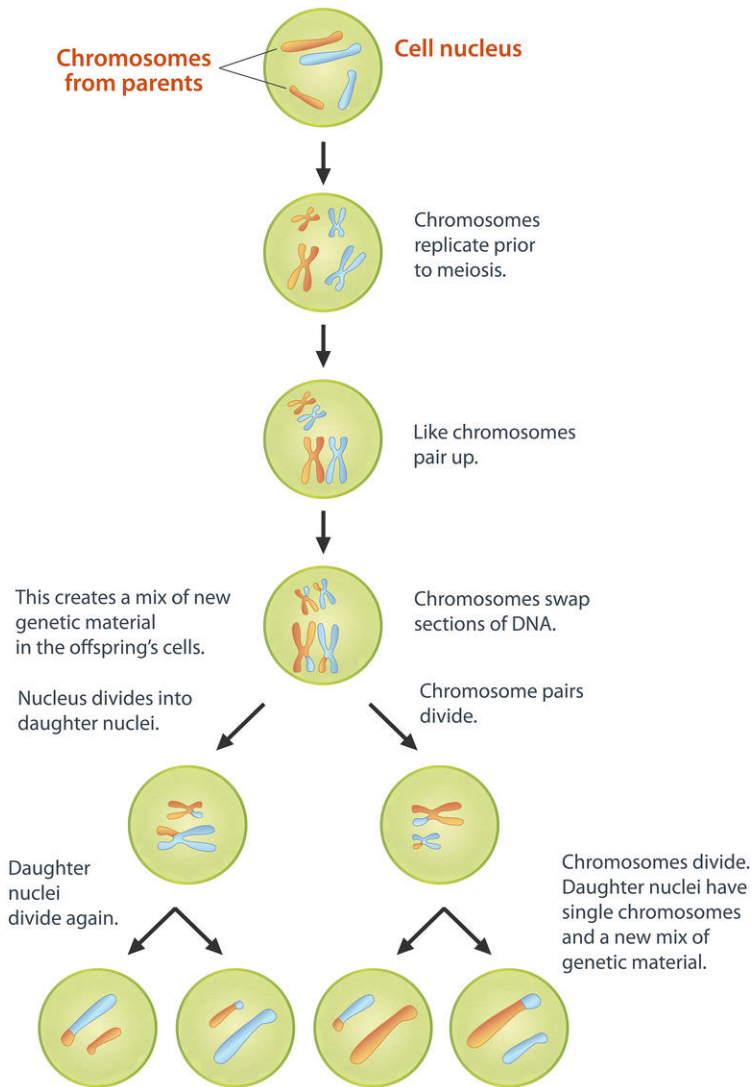
During meiosis II, the sister chromatids are separated and the gametes are generated. This cell division is similar to that of **mitosis**, but results in four genetically unique haploid cells. The steps are outlined below:

1. Prophase II: The chromosomes condense.
2. Metaphase II: The chromosomes line up one on top of each other along the middle of the cell, similar to how they line up in mitosis. The spindle is attached to the centromere of each chromosome.
3. Anaphase II: The sister chromatids separate as the spindle shortens and move to opposite ends of the cell.
4. Telophase II: A nuclear envelope forms around the chromosomes in all four cells. This is followed by cytokinesis.

After cytokinesis, each cell has divided again. Therefore, meiosis results in four haploid genetically unique daughter cells, each with half the DNA of the parent cell (**Figure 10.3**). In human cells, the parent cell has 46 chromosomes (23 pairs), so the cells produced by meiosis have 23 chromosomes. These cells will become gametes.

Summary

- Meiosis is a process of cell division that reduces the chromosome number by half and produces sex cells, or gametes.
- Meiosis is divided into two parts: Meiosis I and Meiosis II. Each part is similar to mitosis and can be divided into the same phases: prophase, metaphase, anaphase, and telophase.
- Crossing-over occurs only during prophase I.
- Four genetically unique haploid cells result from meiosis.

**FIGURE 10.3**

An overview of meiosis.

Explore More

Use the resource below to answer the questions that follow.

- **Meiosis** on YouTube at http://www.youtube.com/watch?v=rB_8dTuh73c (9:15)



MEDIA

Click image to the left or use the URL below.

URL: <https://www.ck12.org/flx/render/embeddedobject/114303>

1. What is meiosis?
2. What is diploid? How many chromosomes are in a diploid human cell?
3. What is a zygote? How does the zygote form the organism?
4. What is the result of crossing-over?
5. How many cell divisions occur during meiosis?
6. Why are you genetically distinct?

Review

1. Define meiosis.
2. What is the difference between a haploid cell and a diploid cell?
3. Describe the steps of Meiosis I and Meiosis II.
4. Describe crossing-over. When does crossing-over occur?
5. What is the outcome of meiosis?

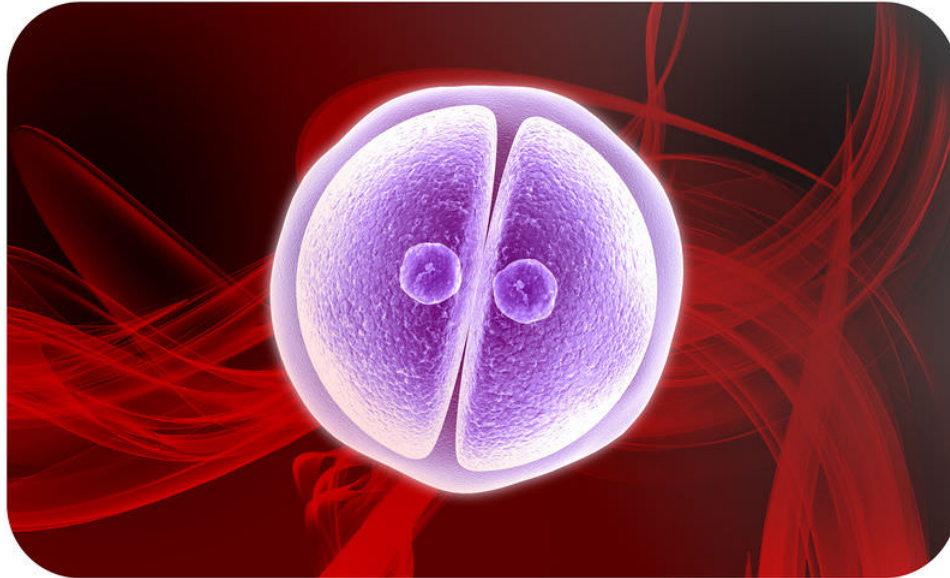
References

1. Hana Zavadska. [Overview of Meiosis](#) . CC BY-NC 3.0
2. Masur. [Diagram of crossing-over](#) . Public Domain
3. Hana Zavadska. [An overview of meiosis](#) . CC BY-NC 3.0

CONCEPT 11

Mitosis vs. Meiosis

- Distinguish between mitosis and meiosis.
- Summarize the necessity for mitosis and meiosis.

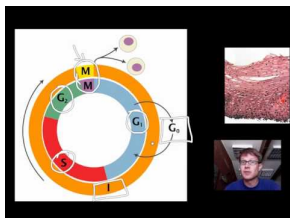


Mitosis or Meiosis?

This represents a tiny embryo just beginning to form. Once an egg is fertilized, the resulting single cell must divide many, many times to develop a fetus. Both mitosis and meiosis involve cell division; is this type of cell division an example of mitosis or meiosis? The answer is mitosis. With each division you are making a genetically exact copy of the parent cell, which only happens through mitosis.

Mitosis vs. Meiosis

Mitosis, meiosis, and sexual reproduction are discussed at <https://www.youtube.com/watch?v=2aVnN4RePyI>.



MEDIA

Click image to the left or use the URL below.

URL: <https://www.ck12.org/flx/render/embeddedobject/180471>

Both **mitosis** and **meiosis** result in eukaryotic cells dividing. So what is the difference between mitosis and meiosis? The primary difference is the differing goals of each process. The goal of mitosis is to produce two **daughter cells** that are genetically identical to the parent cell, meaning the new cells have exactly the same DNA as the parent cell. Mitosis happens when you want to grow, for example. You want all your new cells to have the same DNA as the previous cells. The goal of meiosis, however, is to produce sperm or eggs, also known as **gametes**. The resulting gametes are not genetically identical to the parent cell. Gametes are **haploid** cells, with only half the DNA present

in the **diploid** parent cell. This is necessary so that when a sperm and an egg combine at **fertilization**, the resulting **zygote** has the correct amount of DNA—not twice as much as the parents. The zygote then begins to divide through mitosis.

Pictured below is a comparison between **binary fission** (Figure 11.1), which is cell division of prokaryotic organisms, mitosis, and meiosis. Mitosis and meiosis are also compared in the table that follows (Table 11.1).

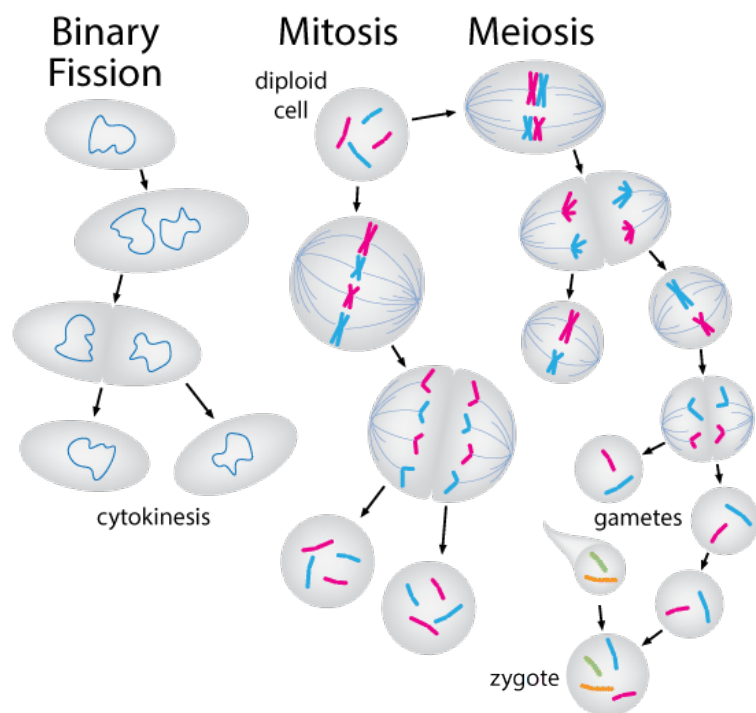


FIGURE 11.1

A comparison between binary fission, mitosis, and meiosis.

TABLE 11.1: Mitosis vs. Meiosis: A Comparison

	Mitosis	Meiosis
Purpose	To produce new cells	To produce gametes
Number of Cells Produced	2	4
Rounds of Cell Division	1	2
Haploid or Diploid	Diploid	Haploid
Daughter cells identical to parent cells?	Yes	No
Daughter cells identical to each other?	Yes	No

Summary

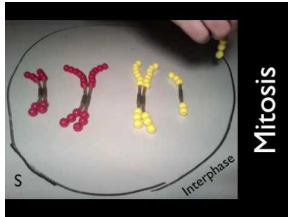
- The goal of mitosis is to produce a new cell that is identical to the parent cell.
- The goal of meiosis is to produce gametes that have half the DNA of the parent cell.

Explore More

Use the resources below to answer the questions that follow.

Explore More I

- **Mitosis and Meiosis Simulation** at <http://www.youtube.com/watch?v=zGVBAHAsjJM> (11:53)



MEDIA

Click image to the left or use the URL below.

URL: <https://www.ck12.org/flx/render/embeddedobject/57344>

1. What are homologous chromosomes?
2. How do the location of specific genes compare between homologous chromosomes?
3. What is the outcome from mitosis?
4. What is a tetrad? Why are they an important feature of meiosis?
5. How does meiosis differ between females and males?

Explore More II

- **How Cells Divide** at <http://www.pbs.org/wgbh/nova/body/how-cells-divide.html>

1. How many daughter cells arise from mitosis? How many daughter cells are produced in meiosis?
2. How does the attachment of spindle fibers differ between mitosis and meiosis I?
3. Is anaphase I or anaphase II in meiosis more analogous to anaphase in mitosis? Explain your reasoning.
4. How many steps are there in mitosis? How many steps are there in meiosis?
5. How does interphase I of meiosis differ from interphase II of meiosis?

Review

1. What is the goal of mitosis? Of meiosis?
2. How many cells are created from cytokinesis following mitosis? Following meiosis?
3. Which process, mitosis to meiosis, creates genetically identical cells?
4. "Gametes are haploid cells." What does this sentence mean?

References

1. Zachary Wilson. [A comparison between binary fission, mitosis, and meiosis](#) . CC BY-NC 3.0

CONCEPT 12

Asexual vs. Sexual Reproduction

- Distinguish between sexual and asexual reproduction.
- Describe the types of asexual reproduction.
- Explain how plants and animals reproduce sexually.



Do animals always have two parents?

No, not all animals have two parents. When necessary, some animals can be produced from just one parent. Some reptiles, such as this Komodo dragon, have only one parent. The process of creating offspring from just one individual is called asexual reproduction.

Reproduction

Animals and other organisms cannot live forever. They must reproduce if their species is to survive. But what does it mean to reproduce? **Reproduction** is the ability to make the next generation, and it is one of the basic characteristics of life. Two methods of reproduction are:

1. **Asexual reproduction**, the process of forming a new individual from a single parent.

2. **Sexual reproduction**, the process of forming a new individual from two parents.

There are advantages and disadvantages to each method, but the result is always the same: a new life begins.

Asexual Reproduction

When humans reproduce, there are two parents involved. DNA must be passed from both the mother and father to the child. Humans cannot reproduce with just one parent; humans can only reproduce sexually. But having just one parent is possible in other eukaryotic organisms, including some insects, fish, and reptiles. These organisms can reproduce asexually, meaning the offspring ("children") have a single parent and share the exact same genetic material as the parent. This is very different from reproduction in humans. Bacteria, being a prokaryotic, single-celled organism, must reproduce asexually.

The advantage of asexual reproduction is that it can be very quick and does not require the meeting of a male and female organism. The disadvantage of asexual reproduction is that organisms do not receive a mix of traits from both parents. An organism that is born through asexual reproduction only has the DNA from the one parent. In fact, the offspring is genetically an exact copy of the parent. This can cause problems for the individual. For example, if the parent has a gene that causes a particular disease, the offspring will also have the gene that causes that disease. Organisms produced sexually may or may not inherit the disease gene because they receive a mix of their parents' genes.

Types of organisms that reproduce asexually include:

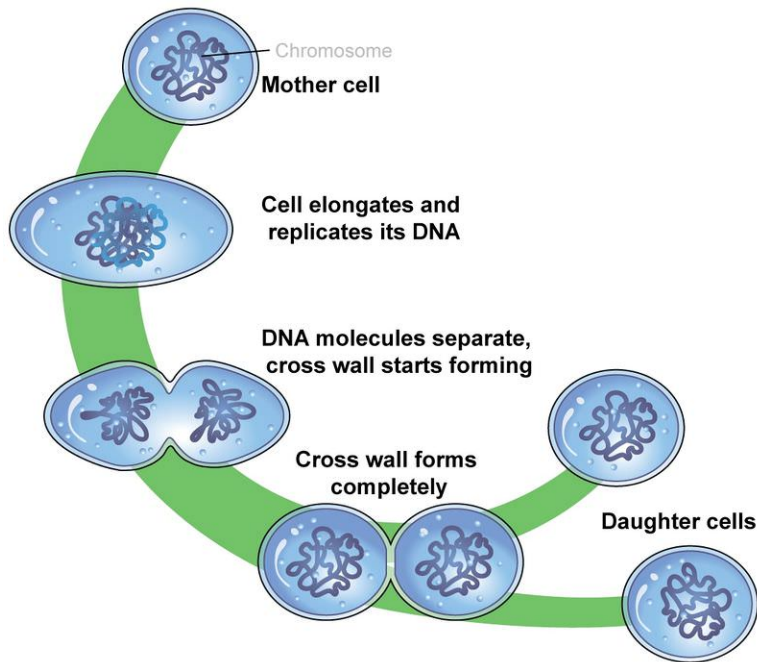
1. Prokaryotic organisms, like bacteria. Bacteria reproduce through **binary fission**, where they grow and divide in half (**Figure 12.1**). First, their chromosome replicates and the cell enlarges. The cell then divides into two cells as new membranes form to separate the two cells. After cell division, the two new cells each have one identical chromosome. This simple process allows bacteria to reproduce very rapidly.
2. Flatworms, an invertebrate animal species. Flatworms divide in two, then each half regenerates into a new flatworm identical to the original, a process called **fragmentation**.
3. Different types of insects, fish, and lizards. These organisms can reproduce asexually through a process called parthenogenesis. **Parthenogenesis** happens when an unfertilized egg cell grows into a new organism. The resulting organism has half the amount of genetic material of the parent. Parthenogenesis is common in honeybees. In a hive, the sexually produced eggs become workers, while the asexually produced eggs become drones.

Sexual Reproduction

During sexual reproduction, two parents are involved. Most animals are **dioecious**, meaning there is a separate male and female sex, with the male producing sperm and the female producing eggs. When a sperm and egg meet during **fertilization**, a **zygote**, the first cell of a new organism, is formed (**Figure 12.2**). This process combines the genetic material from both parents. The resulting organism will be genetically unique. The zygote will divide by mitosis and grow into the embryo.

Let's explore how animals, plants, and fungi reproduce sexually:

Animals often have **gonads**, organs that produce eggs or sperm. The male gonads are the **testes**, and the female gonads are the **ovaries**. Testes produce sperm; ovaries produce eggs. Sperm and egg, the two sex cells, are known as **gametes**, and can combine two different ways, both of which combine the genetic material from the two parents. Gametes have half the amount of the genetic material of a regular body cell; they are **haploid** cells. In humans, gametes have one set of 23 chromosomes. Gametes are produced through a special type of cell division known as **meiosis**. Normal human cells have 46 chromosomes. They are **diploid** cells, with two sets of 23 chromosomes (23 pairs).

**FIGURE 12.1**

Bacteria reproduce by binary fission. Shown is one bacterium reproducing and becoming two bacteria.

**FIGURE 12.2**

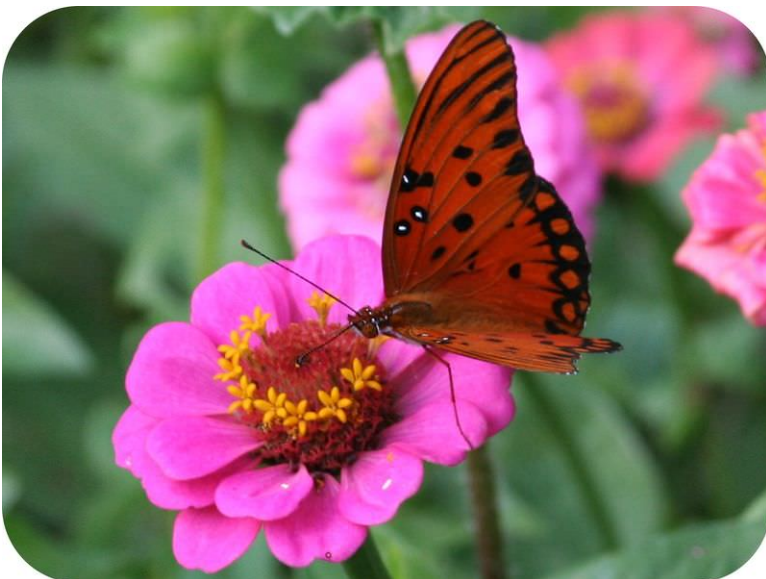
During sexual reproduction, a sperm fertilizes an egg.

Fish and other aquatic animals release their gametes in the water, which is called **external fertilization** (Figure 12.3). These gametes will combine by chance. Animals that live on land (reptiles, birds, and mammals) reproduce by **internal fertilization**. Typically males have a penis that deposits sperm into the vagina of the female. Birds do not have penises, but they do have a chamber called the cloaca that they place close to another bird's cloaca to deposit sperm. Amphibians must live close to water as they must lay their eggs in a moist or wet environment prior to external fertilization.

**FIGURE 12.3**

This fish guards her eggs, which will be fertilized externally.

- Plants can also reproduce sexually, but their reproductive organs are different from animals' gonads. Plants that have flowers have their reproductive parts in the flower. The sperm is contained in the pollen, while the egg is contained in the ovary, deep within the flower. The sperm can reach the egg two different ways:
 1. In **self-pollination**, the egg is fertilized by the pollen of the same flower.
 2. In **cross-pollination**, sperm from the pollen of one flower fertilizes the egg of another flower. Like other types of sexual reproduction, cross-pollination allows new combinations of traits. Cross-pollination occurs when pollen is carried by the wind to another flower. It can also occur when animal pollinators, like honeybees or butterflies (**Figure 12.4**), carry the pollen from flower to flower.

**FIGURE 12.4**

Butterflies receive nectar when they deposit pollen into flowers, resulting in cross-pollination.

- Fungi can also reproduce sexually, but instead of female and male sexes, they have (+) and (-) strains. When the filaments of a (+) and (-) fungi meet, the zygote is formed. Just like in plants and animals, each zygote receives DNA from two parent strains.

Summary

- Types of asexual reproduction, when a new individual is formed from a single parent, include binary fission in bacteria and parthenogenesis in some animals.
- During sexual reproduction in animals, fertilization can be internal or external.
- Cross-pollination allows sexual reproduction in plants.

Explore More

Use the resource below to answer the questions that follow.

- **Plant reproduction: Asexual Reproduction** at <http://www.youtube.com/watch?v=drcnTg7ZCoc> (2:57)



MEDIA

Click image to the left or use the URL below.

URL: <https://www.ck12.org/flx/render/embeddedobject/57350>

1. How does the production of bulbs benefit plants?
2. How can an organism benefit from asexual reproduction?
3. What can be a negative effect of asexual reproduction? Is this more applicable to the individual or the population?

Review

1. What is asexual reproduction?
2. What is the advantage of sexual reproduction?
3. Describe two types of asexual reproduction.
4. What is a zygote?
5. How many chromosomes are in a human zygote? How many chromosomes are in a human gamete?

References

1. LadyofHats. [Diagram of reproduction by binary fission](#) . CC BY-NC 3.0
2. Image copyright James Steidl, 2014. [During sexual reproduction, a sperm fertilizes an egg](#) . Used under license from Shutterstock.com
3. Chika Watanabe. [Fish eggs are fertilized externally](#) . CC BY 2.0
4. James Emery. [Butterflies cross-pollinate flowers](#) . CC BY 2.0

CONCEPT 13

Mendel's Pea Plants

- Summarize the importance of Gregor Mendel to genetics.
- Distinguish between self-pollination and cross-pollination.
- Describe Mendel's first genetics experiments.



Why do you look like your family?

For a long time people understood that traits are passed down through families. The rules of how this worked were unclear, however. The work of Gregor Mendel was crucial in explaining how traits are passed down to each generation.

Mendel's Experiments

What does the word "inherit" mean? You may have inherited something of value from a grandparent or another family member. To **inherit** is to receive something from someone who came before you. You can inherit objects, but you can also inherit traits. For example, you can inherit a parent's eye color, hair color, or even the shape of your nose and ears!

Genetics is the study of inheritance. The field of genetics seeks to explain how traits are passed on from one generation to the next.

In the late 1850s, an Austrian monk named Gregor Mendel (**Figure 13.1**) performed the first genetics experiments. To study genetics, Mendel chose to work with pea plants because they have easily identifiable traits (**Figure 13.2**). For example, pea plants are either tall or short, which is an easy trait to observe. Furthermore, pea plants grow quickly, so he could complete many experiments in a short period of time.

Mendel also used pea plants because they can either **self-pollinate** or be **cross-pollinated**. Self-pollination means that only one flower is involved; the flower's own pollen lands on the female sex organs. Cross pollination is done by hand by moving pollen from one flower to the stigma of another (just like bees do naturally). As a result, one plant's sex cells combine with another plant's sex cells. This is called a "cross." These crosses produce **offspring**


FIGURE 13.1

Gregor Mendel, the "father" of genetics.















Seed		Flower	Pod		Stem	
Form	Cotyledon	Color	Form	Color	Place	Size
						
Round	Yellow	White	Full	Green	Axial pods	Tall
						
Wrinkled	Green	Violet	Constricted	Yellow	Terminal pods	Short
1	2	3	4	5	6	7

FIGURE 13.2

Characteristics of pea plants.

(or "children"), just like when male and female animals mate. Since Mendel could move pollen between plants, he could carefully control and then observe the results of crosses between two different types of plants.

He studied the inheritance patterns for many different traits in peas, including round seeds versus wrinkled seeds, white flowers versus purple flowers, and tall plants versus short plants. Because of his work, Mendel is considered the "Father of Genetics."

Mendel's First Experiment

In one of Mendel's early experiments, he crossed a short plant and a tall plant. What do you predict the offspring of these plants were? Medium-sized plants? Most people during Mendel's time would have said medium-sized. But an unexpected result occurred. Mendel observed that the offspring of this cross (called the **F1 generation**) were all tall plants!

Next, Mendel let the F1 generation self-pollinate. That means the tall plant offspring were crossed with each other. He found that 75% of their offspring (the **F2 generation**) were tall, while 25% were short. Shortness skipped a generation. But why? In all, Mendel studied seven characteristics, with almost 20,000 F2 plants analyzed. All of his results were similar to the first experiment—about three out of every four plants had one trait, while just one out of every four plants had the other.

For example, he crossed purple flowered-plants and white flowered-plants. Do you think the colors blended? No, they did not. Just like the previous experiment, all offspring in this cross (the F1 generation) were one color: purple. In the F2 generation, 75% of plants had purple flowers and 25% had white flowers (**Figure 13.3**). There was no

blending of traits in any of Mendel's experiments.

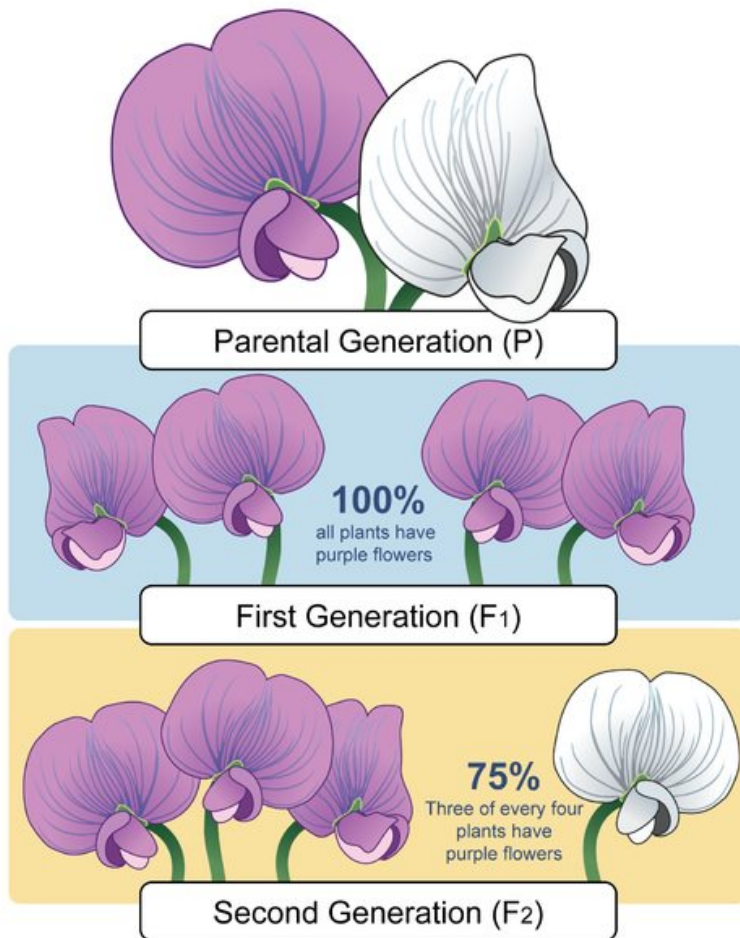


FIGURE 13.3

The results of Mendel's experiment with purple flowered and white flowered-plants numerically matched the results of his experiments with other pea plant traits.

Summary

- Gregor Mendel was the father of the field of genetics, which seeks to explain how traits are passed on from one generation to the next.
- To study genetics, Mendel chose to work with pea plants because they have easily identifiable traits.

Explore More

Use the resource below to answer the questions that follow.

- **Pea experiment** at <http://www.sonic.net/~nbs/projects/anthro201/exper/>

1. What is a "simple" trait?
2. What is a heterozygote? How is this different than a homozygote?
3. You breed a plant with yellow wrinkled peas with a plant with yellow smooth peas. Both individuals are homozygous for both traits. What will the peas of the next generation look like?
4. You breed plants with the same traits as in question 3, but this time the smooth trait is heterozygous in the second individual. What will the peas of the next generation look like?

5. You breed two green wrinkled plants. Will will the next generation look like?

Review

1. What is genetics?
2. Why did Mendel choose to study pea plants?
3. How did Mendel's experiments disprove the idea that we are simply a "blend" of our parents' traits?
4. What were the results Mendel consistently identified in his experiments?

References

1. Erik Nordenskiöld. [Portrait of Gregor Mendel](#) . Public Domain
2. Rupali Raju. [The Laws of Heredity](#) . CC BY-NC 3.0
3. Mariana Ruiz Villarreal (LadyofHats) for CK-12 Foundation. [Result of Mendel's experiment with purple and white flowered plants](#) . CC BY-NC 3.0

CONCEPT **14****Mendel's Laws and Genetics**

- Distinguish between dominant and recessive traits.
- Explain the law of segregation.

**What does it mean to be dominant?**

The most powerful or influential individual in a group is sometimes called dominant. In genetics, a dominant trait means nearly the same thing. A dominant trait is the most influential trait and masks the other trait.

Dominance

Do you remember what happened when Mendel crossed purple flowered-plants and white flowered-plants? All the offspring had purple flowers. There was no blending of traits in any of Mendel's experiments. Mendel had to come up with a theory of inheritance to explain his results. He developed a theory called the **law of segregation**.

The Law of Segregation

Mendel proposed that each pea plant had two hereditary factors for each trait. There were two possibilities for each hereditary factor, such as a purple factor or white factor. One factor is **dominant** to the other. The other trait that is masked is called the **recessive** factor, meaning that when both factors are present, only the effects of the dominant factor are noticeable (**Figure 14.1**). Although you have two hereditary factors for each trait, each parent can only pass on one of these factors to the offspring. When the sex cells, or **gametes** (sperm or egg), form, the heredity factors must separate, so there is only one factor per gamete. In other words, the factors are "segregated" in each gamete. Mendel's law of segregation states that the two hereditary factors separate when gametes are formed. When fertilization occurs, the offspring receive one hereditary factor from each gamete, so the resulting offspring have two factors.

The law of segregation predates our understanding of meiosis. Mendel developed his theories without an understanding of DNA, or even the knowledge that DNA existed. Quite a remarkable feat!



FIGURE 14.1

In peas, purple flowers are dominant to white. If one of these purple flowers is crossed with a white flower, all the offspring will have purple flowers.

Example Cross

This law explains what Mendel had seen in the F1 generation when a tall plant was crossed with a short plant. The two heredity factors in this case were the short and tall factors. Each individual in the F1 would have one of each factor, and as the tall factor is dominant to the short factor (the recessive factor), all the plants appeared tall.

In describing genetic crosses, letters are used. The dominant factor is represented with a capital letter (T for tall) while the recessive factor is represented by a lowercase letter (t). For the T and t factors, three combinations are possible: TT , Tt , and tt . TT plants will be tall, while plants with tt will be short. Since T is dominant to t , plants that are Tt will be tall because the dominant factor masks the recessive factor.

In this example, we are crossing a TT tall plant with a tt short plant. As each parent gives one factor to the F1 generation, all of the F1 generation will be Tt tall plants.

When the F1 generation (Tt) is allowed to self-pollinate, each parent will give one factor (T or t) to the F2 generation. So the F2 offspring will have four possible combinations of factors: TT , Tt , tT , or tt . According to the laws of probability, 25% of the offspring would be tt , so they would appear short. And 75% would have at least one T factor and would be tall.

Summary

- One hereditary factor is dominant to the other. The dominant trait masks the recessive factor, so that when both factors are present, only the effects of the dominant factor are noticeable.
- According to Mendel's law of segregation, there are two hereditary factors for each trait that must segregate during gamete (egg and sperm) production. As a result, offspring receive one factor from each parent, resulting in two factors for each trait in the offspring.

Explore More

Use the resource below to answer the questions that follow.

- **Mendel's Experiment** at <http://www.sumanasinc.com/webcontent/animations/content/mendel/mendel.html>

1. In Mendel's experiments, did it matter if the dominant trait came from the seed plant or the pollen plant?
2. Yellow is a dominant trait in peas. You breed two plants with yellow peas, and some of the offspring's peas are green? How can this be? Explain your answer fully.
3. For some of his experiments Mendel saw a 9:3:3:1 ratio, consisting of 9 yellow/smooth, 3 yellow/wrinkled, 3 green/smooth, and 1 green/wrinkled. What did he conclude from this ratio? Explain where these ratios came from.

Review

1. What is the difference between a dominant trait and a recessive trait?
2. Explain the law of segregation.
3. When Mendel crossed a TT tall plant with a tt short plant, what did he observe in the F1 generation? Why?
4. If PP purple plants are crossed with pp white plants, what will be the possible combinations of factors if the F1 generation is allowed to self-pollinate?

References

1. Forest and Kim Starr/Starr Environmental. [Purple flowers are dominant to white in pea plants](#) . CC BY 2.0

CONCEPT **15**

Punnett Squares

- Explain the relationship between probability and genetics.
- Use a Punnett square to make predictions about the traits of the offspring of a genetic cross.



What's the chance of the coin landing on heads?

There is always a 50-50 chance that a coin will land on heads. Half the time it will land on heads and half the time it will land on tails. What is the chance of it landing heads twice in a row? Or three times? These rules of probability also apply to genetics.

A parent only gives one factor for each trait to an offspring. If a parent has one dominant and one recessive factor for a trait, then, on average, half the time the dominant factor will be passed on, and half the time the recessive factor will be passed on.

Probability and Punnett Squares

A **Punnett square** is a special tool derived from the laws of probability. It is used to predict the possible offspring from a cross, or mating between two parents.

An example of a Punnett square (**Figure 15.1**) shows the results of a cross between two purple flowers that each have one dominant factor and one recessive factor (Bb).

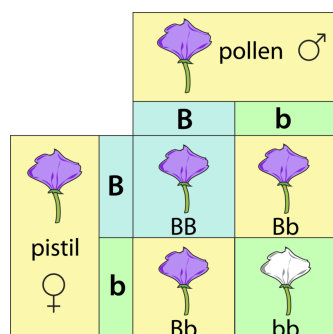


FIGURE 15.1

The Punnett square of a cross between two purple flowers (Bb). A Punnett square can be used to calculate what percentage of offspring will have a certain trait.

To create a Punnett square, perform the following steps:

1. Take the factors from the first parent and place them at the top of the square (B and b).
2. Take the factors from the second parent and line them up on the left side of the square (B and b).
3. Pull the factors from the top into the boxes below.
4. Pull the factors from the side into the boxes next to them.

The possible offspring are represented by the letters in the boxes, with one factor coming from each parent.

Results:

- Top left box: BB , or purple flowers
- Top right box: Bb , or purple flowers
- Lower left box: Bb , or purple flowers
- Lower right box: bb , or white flowers

Only one of the plants out of the four, or 25% of the plants, has white flowers (bb). The other 75% have purple flowers (BB , Bb), because the purple factor (B) is the dominant factor. This shows that the color purple is the **dominant trait** in pea plants.

Now imagine you cross one of the white flowers (bb) with a purple flower that has both a dominant and recessive factor (Bb). The only possible gamete in the white flower is recessive (b), while the purple flower can have gametes with either dominant (B) or recessive (b).

Practice using a Punnett square with this cross (see **Table 15.1**).

TABLE 15.1: White Flower (bb) Crossed with Purple Flower (Bb)

	b	b
B	Bb	Bb
b	bb	bb

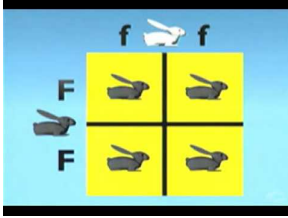
Did you find that 50% of the offspring will be purple, and 50% of the offspring will be white?

Summary

- A Punnett square is a special tool used to predict the offspring from a cross, or mating between two parents.
- In a Punnett square, the possible offspring are represented by the letters in the boxes, with one factor coming from each parent.

Explore More

Use the resources below to answer the questions that follow.

**MEDIA**

Click image to the left or use the URL below.

URL: <https://www.ck12.org/flx/render/embeddedobject/57307>

1. What are Mendel's two rules of heredity?
2. If a pure bred black rabbit and a white rabbit mate, what will their potential offspring look like?
3. If two hybrid Ff rabbits mate, what will the offspring look like?

Explore More II

- **Punnett Square Calculator** at <http://scienceprimer.com/punnett-square-calculator>

1. If you cross an Aa individual with another Aa individual, what will the genotype ratio be in the next generation? What will be the phenotype ratio?
2. If you cross an $AABb$ individual with an $Aabb$ individual, what will the genotype ratio be in the next generation? What will be the phenotype ratio?
3. If you cross an $AABb$ individual with an $aabb$ individual, what will the genotype ratio be in the next generation? What will be the phenotype ratio?

Review

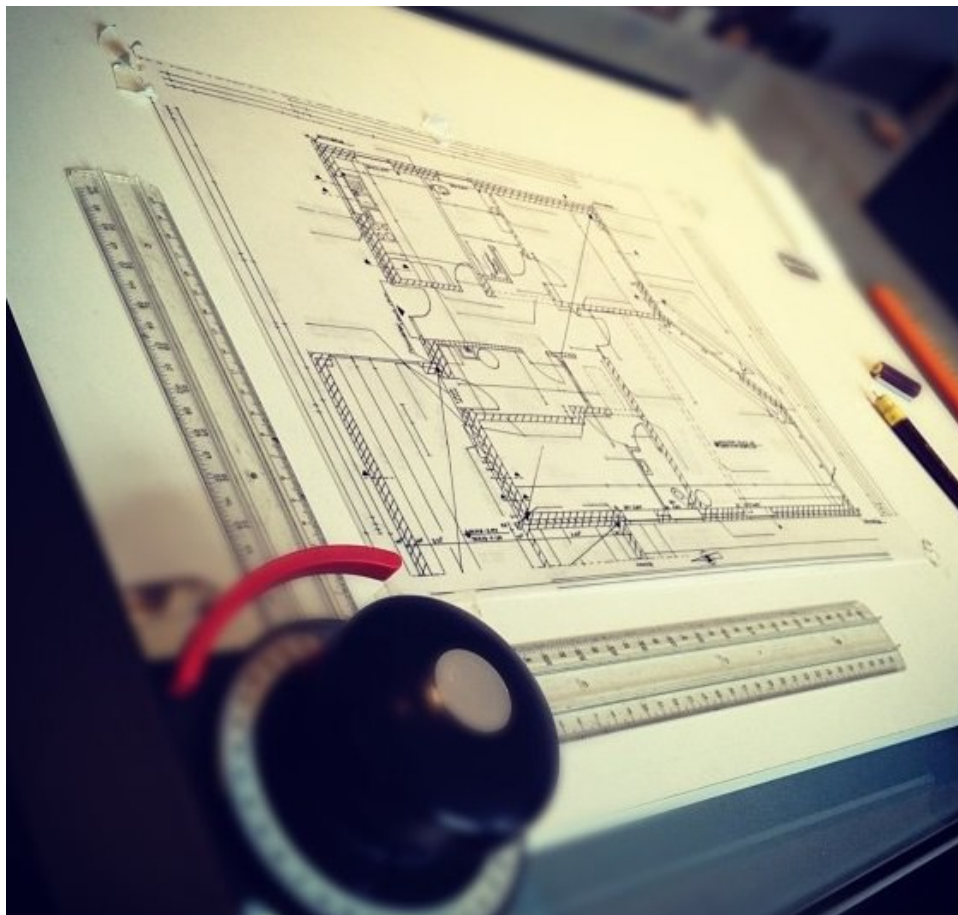
1. In peas, yellow seeds (Y) are dominant over green seeds (y). If a yy plant is crossed with a YY plant, what ratio of plants in the offspring would you predict?
2. What ratio of plants in the offspring would you predict from a $Yy \times Yy$ cross?
3. In guinea pigs, smooth coat (S) is dominant over rough coat (s). If an SS guinea pig is crossed with an ss guinea pig, what ratio of guinea pigs in the offspring would you predict?
4. What ratio of guinea pigs in the offspring would you predict from a $Ss \times ss$ cross?

References

1. Jodi So. [The Punnett square of a cross between two heterozygous purple flowers](#) .

CONCEPT 16 DNA, the Genetic Material

- Explain the importance of DNA.
- Define and describe a nucleotide.
- Describe the shape of DNA.
- Summarize the base-pairing rules.



Where's the instructions?

How do your cells know what to do? Just like builders have blueprints to tell them how to build a house, your cells also have instructions. Your cells' instructions are molecules of DNA.

What is DNA?

DNA is the material that makes up our chromosomes and stores our genetic information. When you build a house, you need a blueprint, a set of instructions that tells you how to build. The DNA is like the blueprint for living organisms. The genetic information is a set of instructions that tell your cells what to do.

DNA is an abbreviation for **deoxyribonucleic acid**. As you may recall, **nucleic acids** are a type of macromolecule that store information. The *deoxyribo* part of the name refers to the name of the sugar that is contained in DNA, deoxyribose. DNA may provide the instructions to make up all living things, but it is actually a very simple molecule. DNA is made of a very long chain of nucleotides. In fact, in you, the smallest DNA molecule has well over 20 million nucleotides.

Nucleotides

Nucleotides are composed of three main parts:

1. a phosphate group.
2. a 5-carbon sugar (deoxyribose in DNA).
3. a nitrogen-containing base.

The only difference between each nucleotide is the identity of the base. There are only four possible bases that make up each DNA nucleotide: adenine (A), guanine (G), thymine (T), and cytosine (C).

The Genetic Code

The various sequences of the four nucleotide bases make up the genetic code of your cells. It may seem strange that there are only four letters in the “alphabet” of DNA. But since your **chromosomes** contain millions of nucleotides, there are many, many different combinations possible with those four letters.

But how do all these pieces fit together? James Watson and Francis Crick won the Nobel Prize in 1962 for piecing together the structure of DNA. Together with the work of Rosalind Franklin and Maurice Wilkins, they determined that DNA is made of two strands of nucleotides formed into a **double helix**, or a two-stranded spiral, with the sugar and phosphate groups on the outside, and the paired bases connecting the two strands on the inside of the helix (**Figure 16.1**).

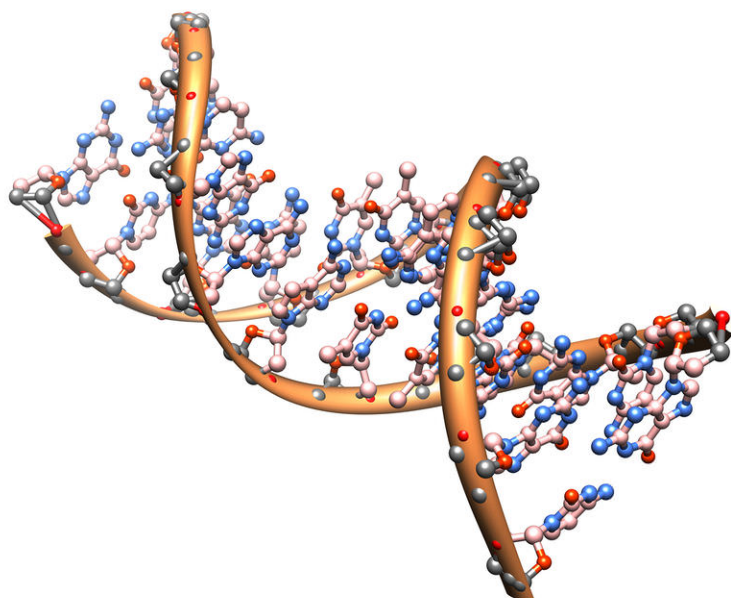


FIGURE 16.1

DNA's three-dimensional structure is a double helix. The hydrogen bonds between the bases at the center of the helix hold the helix together.

Base-Pairing

The bases in DNA do not pair randomly. When Erwin Chargaff looked closely at the bases in DNA, he noticed that the percentage of adenine (A) in the DNA always equaled the percentage of thymine (T), and the percentage of guanine (G) always equaled the percentage of cytosine (C). Watson and Crick's model explained this result by suggesting that A always pairs with T, and G always pairs with C in the DNA helix. Therefore A and T, and G and C, are "complementary bases," or bases that always pair together, known as a **base-pair**. The base-pairing rules

state that A will always bind to T, and G will always bind to C (**Figure 16.2**). For example, if one DNA strand reads ATGCCAGT, the other strand will be made up of the complementary bases: TACGGTCA.

Hydrogen bonds hold the complementary bases together, with two bonds forming between an A and a T, and three bonds between a G and a C.

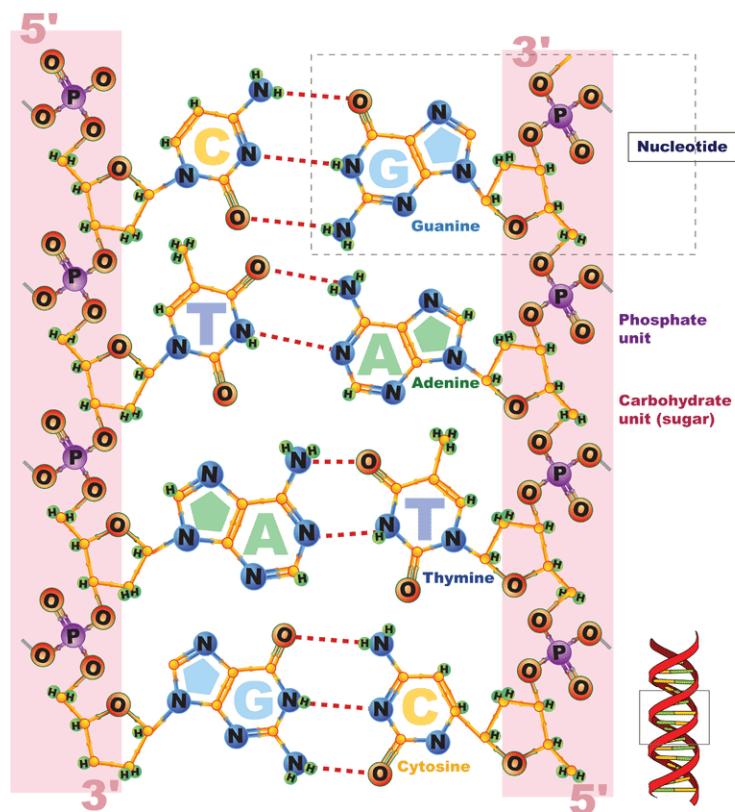


FIGURE 16.2

The chemical structure of DNA includes a chain of nucleotides consisting of a 5-carbon sugar, a phosphate group, and a nitrogen base. Notice how the sugar and phosphate group form the backbone of DNA (strands highlighted in pink), with the hydrogen bonds between the bases joining the two strands.

Summary

- DNA stores the genetic information of the cell in the sequence of its 4 bases: adenine, thymine, guanine, and cytosine.
- DNA is made of a long chain of nucleotides consisting of a 5-carbon sugar, a phosphate group, and nitrogen-containing base.

Explore More

Use the resources below to answer the questions that follow.

Explore More I

- **What Is DNA?** at <http://learn.genetics.utah.edu/content/begin/dna/> . Click on the box "What Is DNA?" on the left side of the page.

1. Where is nuclear DNA located in a eukaryotic organism?

2. Describe the structure of DNA.
3. What is another name for the "sentences" that DNA encodes?
4. What do genes tell a cell to do?

Explore More II

- Go to this link to build a DNA molecule: <http://learn.genetics.utah.edu/content/begin/dna/builddna/> .
1. How long does it take a human cell to copy the DNA in its nucleus before it divides?
 2. How many new cells does your body produce every day?
 3. How many hydrogen bonds form between guanine and cytosine? How many hydrogen bonds form between adenine and thymine? Do you think this relationship helps minimize errors? Explain your reasoning.
 4. What steps does the cell take to speed the rate of DNA replication?

Review

1. Describe the structure of DNA.
2. What does a nucleotide consist of?
3. What are the base-pairing rules?
4. If one DNA strand reads CCGTAATGCAT, what will be the sequence of the complementary strand?

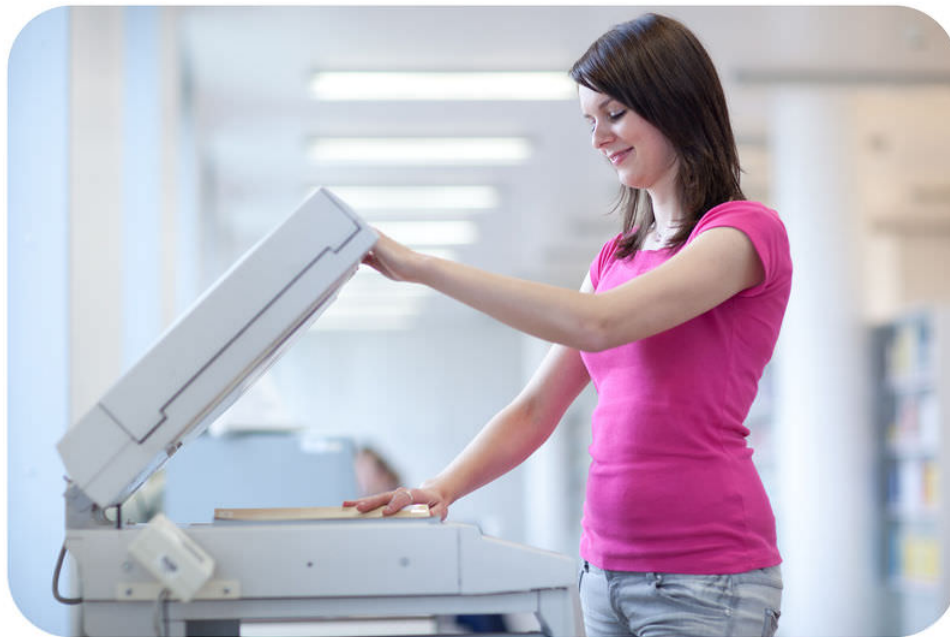
References

1. Image copyright ermess, 2014. [The double helix of DNA](#) . Used under license from Shutterstock.com
2. Mariana Ruiz Villarreal (LadyofHats). [The chemical structure of DNA](#) .

CONCEPT 17

DNA Structure and Replication

- Explain why DNA must replicate itself.
- Describe the process of DNA replication.
- Explain the meaning of semiconservative replication.



Does DNA copy itself?

Yes, your DNA needs to copy itself every time a new cell is created. The new cell needs to have DNA exactly like the rest of your cells. Otherwise, that cell might malfunction. That's why it's important that the process of copying DNA, called DNA replication, is very accurate.

DNA Replication

DNA must replicate (copy) itself so that each resulting cell after mitosis and cell division has the same DNA as the parent cell. All these cells, the parent cell and the two new daughter cells, are genetically identical.

DNA replication occurs during the S phase (the Synthesis phase) of the cell cycle, before mitosis and cell division. The base pairing rules are crucial for the process of replication. DNA replication occurs when DNA is copied to form an identical molecule of DNA.

The general steps involved in DNA replication are as follows:

1. The DNA helix unwinds like a zipper as the bonds between the base pairs are broken. The enzyme DNA Helicase is involved in breaking these bonds.
2. The two single strands of DNA then each serve as a template for a new strand to be created. Using DNA as a template means that on the new strand, the bases are placed in the correct order because of the base pairing rules. Recall that A and T are complementary bases, as are G and C. As a template strand is read, the new strand is created. If ATGCCA is on the "template strand," then TACGGT will be on the new DNA strand. The enzyme DNA Polymerase reads the template and builds the new strand of DNA.

- The new set of nucleotides then join together to form a new strand of DNA. The process results in two DNA molecules, each with one old strand and one new strand of DNA.

This process is known as **semiconservative replication** because one strand is conserved (kept the same) in each new DNA molecule (**Figure 17.1**).

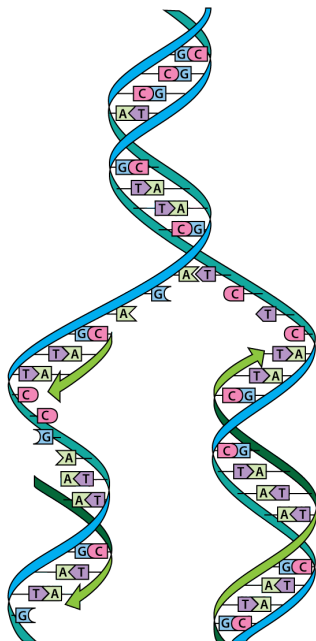


FIGURE 17.1

DNA replication occurs when the DNA strands “unzip,” and the original strands of DNA serve as a template for new nucleotides to join and form a new strand.

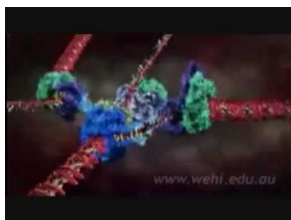
Summary

- During DNA replication, the DNA helix unwinds and the two single strands of DNA then each serve as a template for a new stand to be created.
- DNA replication is semi-conservative: the new DNA molecule consists of half of the parent DNA molecule.

Explore More

Use the resource below to answer the questions that follow.

- DNA replication** at http://www.youtube.com/watch?v=yqESR7E4b_8 (7:47)



MEDIA

Click image to the left or use the URL below.

URL: <https://www.ck12.org/flx/render/embeddedobject/57304>

- What protein molecules does DNA wrap around to form a nucleosome?

2. What makes up chromatin?
3. When can you see chromosomes in a cell?
4. Are both strands of DNA copied continuously during replication? Explain your answer.

Review

1. Describe how DNA is replicated.
2. Explain why DNA replication is sometimes called semiconservative.

References

1. Zachary Wilson. [Diagram of DNA replication](#) . CC BY-NC 3.0

CONCEPT 18**Mutations**

- Define mutation.
- Distinguish between point mutations and chromosomal mutations.
- Explain the outcome from a frameshift mutation.
- Describe the types of chromosomal mutations.
- Explain how mutations occur.

**Would a mutation make you a superhero?**

In the comic books, a mutation can give a person superpowers. Do you think this really happens? In real life, a mutation can be beneficial, or it can harm an organism. For example, beneficial mutations lead to evolution, and harmful mutations can lead to diseases like cancer. A mutation, however, is not going to turn you into a superhero!

Mutations

The process of DNA replication is not always 100% accurate. Sometimes the wrong base is inserted in the new strand of DNA. This wrong base could become permanent. A permanent change in the sequence of DNA is known

as a **mutation**. Small changes in the DNA sequence are usually **point mutations**, which is a change in a single nucleotide. Once DNA has a mutation, that mutation will be copied each time the DNA replicates. After cell division, each resulting cell will carry the mutation.

A mutation may have no effect. However, sometimes a mutation can cause a protein to be made incorrectly. A defect in the protein can affect how well the protein works, or whether it works at all. Usually the loss of a protein function is detrimental to the organism.

In rare circumstances, though, the mutation can be beneficial. Mutations are a mechanism for how species evolve. For example, suppose a mutation in an animal's DNA causes the loss of an enzyme that makes a dark pigment in the animal's skin. If the population of animals has moved to a light colored environment, the animals with the mutant gene would have a lighter skin color and be better camouflaged. So in this case, the mutation is beneficial.

Point Mutations

If a single base is deleted (called a deletion, which is also a point mutation), there can be huge effects on the organism, because this may cause a **frameshift mutation**. Remember that the bases in the mRNA are read in groups of three by the tRNA. If the reading frame is off by even one base, the resulting sequence will consist of an entirely different set of codons.

The reading of an mRNA is like reading three-letter words of a sentence. Imagine the sentence: "The big dog ate the red cat." If you take out the second letter from "big," the frame will be shifted so now it will read: "The bgd oga tet her edc at." One single deletion makes the whole "sentence" impossible to read. A point mutation that adds a base (known as an insertion) would also result in a frameshift.

Chromosomal Mutations

Mutations may also occur in chromosomes (**Figure 18.1**). These mutations are going to be fairly large mutations, possible affecting many genes. Possible types of mutations in chromosomes include:

1. Deletion: When a segment of DNA is lost, so there is a missing segment in the chromosome. These usually result in many genes missing from the chromosome.
2. Duplication: When a segment of DNA is repeated, creating a longer chromosome. These usually result in multiple copies of genes in the chromosome.
3. Inversion: When a segment of DNA is flipped and then reattached to the same chromosome.
4. Insertion: When a segment of DNA from one chromosome is added to another, unrelated chromosome.
5. Translocation: When two segments from different chromosomes change positions.

Causes of Mutations

Many mutations are not caused by errors in replication. Mutations can happen spontaneously, and they can be caused by **mutagens** in the environment. Some chemicals, such as those found in tobacco smoke, can be mutagens. Sometimes mutagens can also cause cancer. Tobacco smoke, for example, is often linked to lung cancer.

Summary

- A mutation is a permanent change in the sequence of bases in DNA.
- Mutations occur in the DNA through deletion, duplication, inversion, insertion, and translocation within the chromosome.
- Mutations can occur due to errors during DNA replication or by mutagens in the environment.

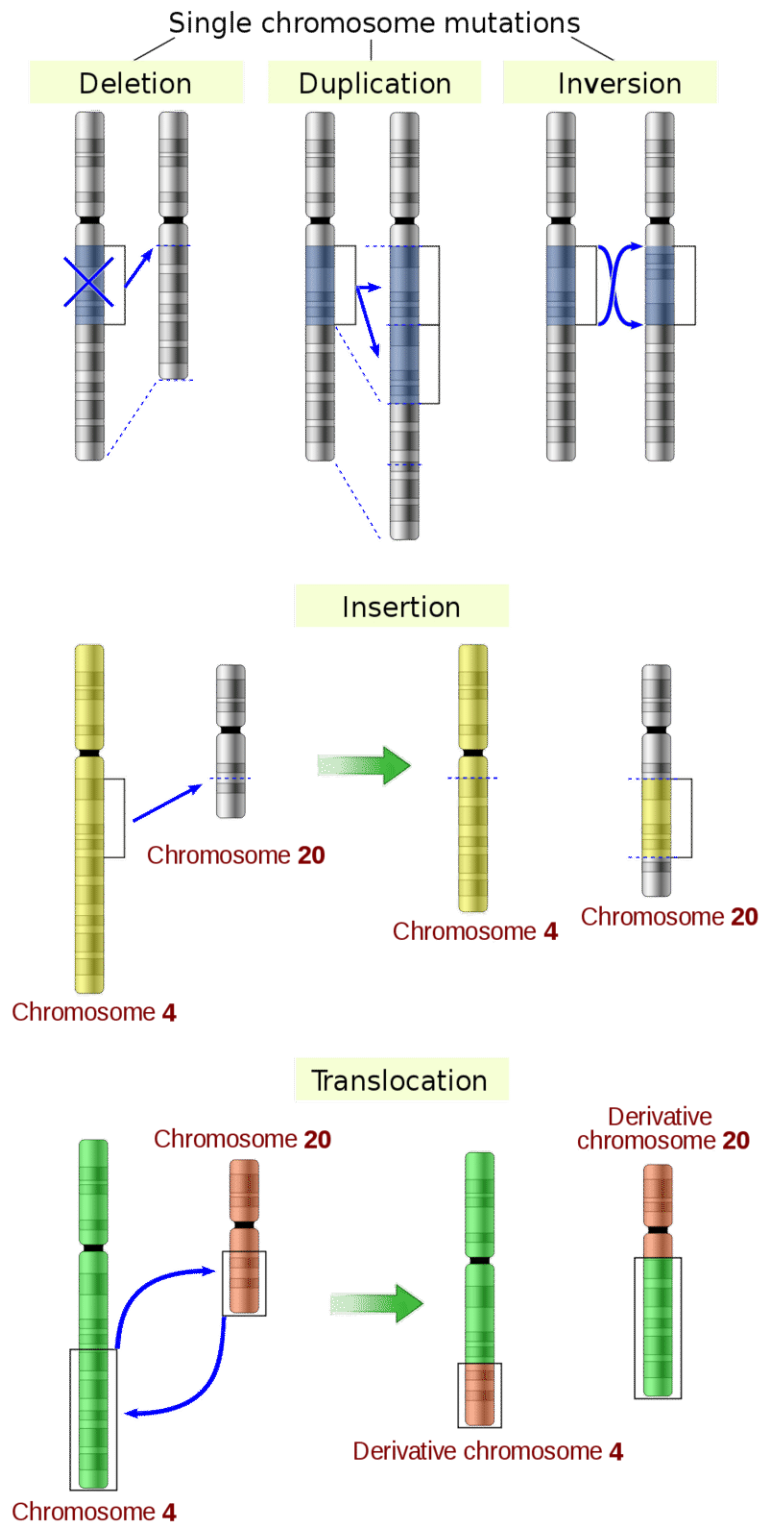


FIGURE 18.1

Mutations can arise in DNA through deletion, duplication, inversion, insertion, and translocation within the chromosome.

Explore More

Use the resources below to answer the questions that follow.

Explore More I

- **Types of Mutations - Understanding Evolution** at http://evolution.berkeley.edu/evolibrary/article/mutations_03
1. What is an example of a genetic disorder caused by a substitution mutation?
 2. How can a substitution mutation change a protein?
 3. Explain a frameshift mutation.
 4. What can cause a frameshift mutation?

Explore More II

- **Gene Regulation** at http://www.teachersdomain.org/asset/novat10_int_evodevo/

Go to this link to see how mutations affect gene regulation. Make sure you make more than one animal and see the effects of more than one mutation occurring at a time.

1. How do mutations in a part of DNA not associated with a gene generally affect the expression of that gene?
2. What do transcription factor proteins do in an organism?

Review

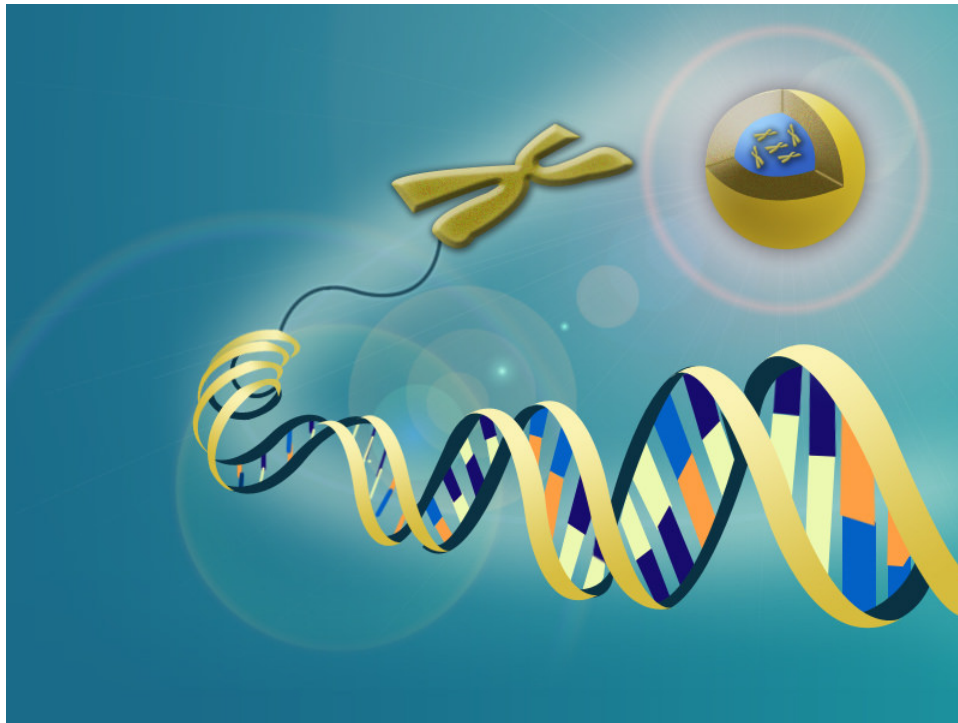
1. Are mutations typically beneficial to the organism?
2. What can cause DNA to mutate?
3. What is a frameshift mutation?
4. Describe two types of chromosomal mutations.

References

1. Yassine Mrabet. [Mutations in chromosomes include deletion, duplication, inversion, insertion, and translocation](#) . Public Domain

CONCEPT **19****Modern Genetics**

- Define allele.
- Compare heterozygous to homozygous.
- Distinguish genotype from phenotype.
- Compare Mendel's laws with the modern understanding of chromosomes.

**Did Mendel know about DNA?**

No, people did not understand that DNA is our hereditary material until long after Mendel's time. Our modern understanding of DNA and chromosomes helped to explain how Mendel's rules worked.

Modern Genetics

Mendel laid the foundation for modern genetics, but there were still a lot of questions he left unanswered. What exactly are the dominant and recessive factors that determine how all organisms look? And how do these factors work?

Since Mendel's time, scientists have discovered the answers to these questions. Genetic material is made out of **DNA**. It is the DNA that makes up the hereditary factors that Mendel identified. By applying our modern knowledge of DNA and chromosomes, we can explain Mendel's findings and build on them. In this concept, we will explore the connections between Mendel's work and modern genetics.

Traits, Genes, and Alleles

Recall that our DNA is wound into **chromosomes**. Each of our chromosomes contains a long chain of DNA that encodes hundreds, if not thousands, of genes. Each of these genes can have slightly different versions from individual

to individual. These variants of genes are called **alleles**. Each parent only donates one allele for each gene to an offspring.

For example, remember that for the height gene in pea plants there are two possible factors. These factors are alleles. There is a dominant allele for tallness (T) and a recessive allele for shortness (t).

Genotype and Phenotype

Genotype is a way to describe the combination of alleles that an individual has for a certain gene (**Table 19.1**). For each gene, an organism has two alleles, one on each chromosome of a homologous pair of chromosomes (think of it as one allele from Mom, one allele from Dad). The genotype is represented by letter combinations, such as TT , Tt , and tt .

When an organism has two of the same alleles for a specific gene, it is **homozygous** (*homo* means "same") for that gene. An organism can be either homozygous dominant (TT) or homozygous recessive (tt). If an organism has two different alleles (Tt) for a certain gene, it is known as **heterozygous** (*hetero* means different).

TABLE 19.1: Genotypes

Genotype	Definition	Example
Homozygous	Two of the same allele	TT or tt
Heterozygous	One dominant allele and one recessive allele	Tt
Homozygous dominant	Two dominant alleles	TT
Homozygous recessive	Two recessive alleles	tt

Phenotype is a way to describe the traits you can see. The genotype is like a recipe for a cake, while the phenotype is like the cake made from the recipe. The genotype expresses the phenotype. For example, the phenotypes of Mendel's pea plants were either tall or short, or they were purple-flowered or white-flowered.

Can organisms with different genotypes have the same phenotypes? Let's see.

What is the phenotype of a pea plant that is homozygous dominant (TT) for the tall trait? Tall. What is the phenotype of a pea plant that is heterozygous (Tt)? It is also tall. The answer is yes, two different genotypes can result in the same phenotype. Remember, the recessive phenotype will be expressed only when the dominant allele is absent, or when an individual is homozygous recessive (tt) (**Figure 19.1**).

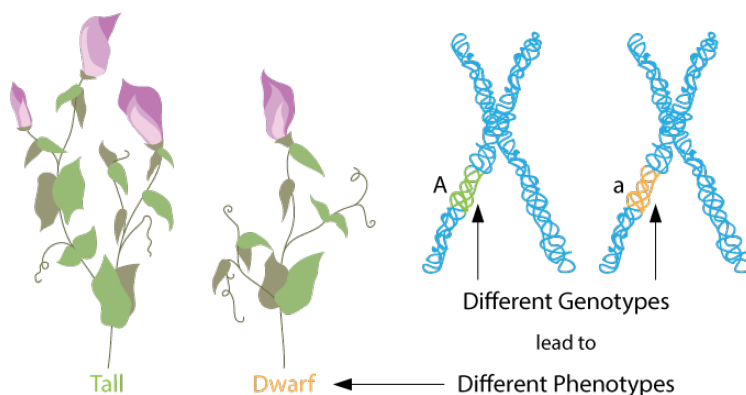


FIGURE 19.1

Different genotypes (AA , Aa , aa or TT , Tt , tt) will lead to different phenotypes, or different appearances of the organism.

Summary

- Mendel's hereditary "factors" are variants of genes called alleles.
- Genotype describes the combination of alleles that an individual has for a certain gene, while phenotype describes the traits that you can see.

Explore More

Use the resources below to answer the questions that follow.

Explore More I

- **Link Between Genotype and Phenotype** at <http://www.sciencelearn.org.nz/Contexts/Uniquely-Me/Sci-Media/Video/Researching-the-link-between-genotype-and-phenotype>
1. When geneticists look at genotype, what are they really studying?
 2. Why do geneticists like to turn genes off? What question(s) do they ask?

Explore More II

- **iPlant Genotype to Phenotype** at http://www.youtube.com/watch?v=nIh0Qy_CZsU (3:49)



MEDIA

Click image to the left or use the URL below.

URL: <https://www.ck12.org/flx/render/embeddedobject/57306>

1. Do most of the complex phenotypes we observe come from a single gene?
2. What has led to the rapid analysis of DNA? Where do scientists now hope to apply these tools?
3. What are some of the phenotypic plant traits that scientists are investigating? Why do you think these traits were chosen?

Review

1. What is an allele?
2. What is the type of allele that only affects the phenotype in the homozygous condition?
3. If two individuals have a certain phenotype, does that mean they must have the same genotype?
4. A tall, green plant is homozygous for each trait. If T is the tall allele, and G is the green allele, what is the genotype and the phenotype of this plant?

References

1. Zachary Wilson. [Different genotypes will lead to different phenotypes of an organism](#) . CC BY-NC 3.0

CONCEPT 20

Polygenic Traits

- Explain the inheritance of polygenic traits.
- Describe the phenotypic distribution of polygenic traits.



Are all people either short or tall?

Unlike Mendel's peas, people do not all fall into two categories: short or tall. Most people, in fact, are somewhere in between. Obviously, Mendel's rules are too simple to explain the inheritance of human height.

Polygenic Traits

Another exception to Mendel's rules is **polygenic inheritance**, which occurs when a trait is controlled by more than one gene. This means that each dominant allele "adds" to the expression of the next dominant allele.

Usually, traits are polygenic when there is wide variation in the trait. For example, humans can be many different sizes. Height is a polygenic trait, controlled by at least three genes with six alleles. If you are dominant for all of the alleles for height, then you will be very tall. There is also a wide range of skin color across people. Skin color is also a polygenic trait, as are hair and eye color.

Polygenic inheritance often results in a bell shaped curve when you analyze the population (**Figure 20.1**). That means that most people fall in the middle of the phenotypic range, such as average height, while very few people are at the extremes, such as very tall or very short. At one end of the curve will be individuals who are recessive for all the alleles (for example, *aabbcc*); at the other end will be individuals who are dominant for all the alleles (for example, *AABBCC*). Through the middle of the curve will be individuals who have a combination of dominant and recessive alleles (for example, *AaBbCc* or *AaBBcc*).

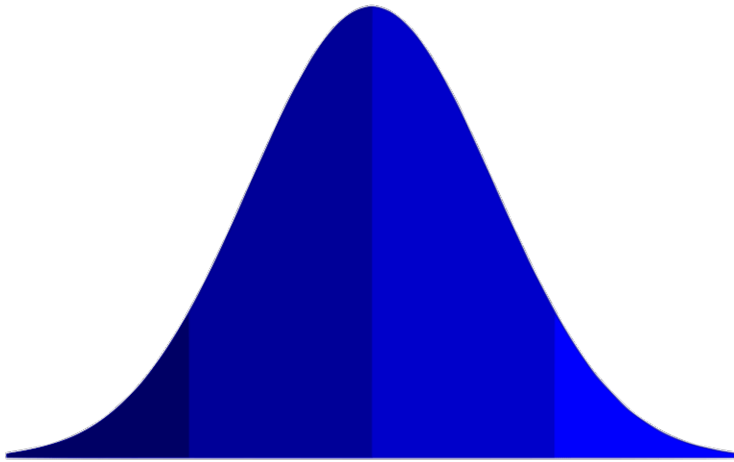


FIGURE 20.1

Polygenic traits tend to result in a distribution that resembles a bell-shaped curve, with few at the extremes and most in the middle. There may be 4 or 6 or more alleles involved in the phenotype. At the left extreme, individuals are completely dominant for all alleles, and at the right extreme, individuals are completely recessive for all alleles. Individuals in the middle have various combinations of recessive and dominant alleles.

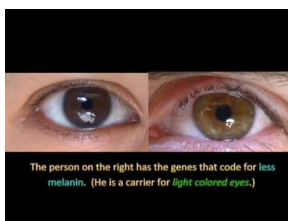
Summary

- In polygenic inheritance, a trait is controlled by more than one gene.
- Examples of polygenic inheritance include height or skin color.

Explore More

Use the resource below to answer the questions that follow.

- **Genetics and Eye Color** at <http://www.youtube.com/watch?v=MjBZaed9yzM> (1:49)



MEDIA

Click image to the left or use the URL below.

URL: <https://www.ck12.org/flx/render/embeddedobject/57303>

1. Is eye color a trait controlled by a single gene as it is often taught in schools?
2. Do you think skin color is a polygenic trait? Explain your reasoning, and be as specific as possible.
3. What is an albino? What kind of eyes would they definitely NOT have?
4. What is known about the melanin levels in people with blue eyes?

Review

1. How does polygenic inheritance violate Mendel's rules?
2. Give examples of traits governed by polygenic inheritance.

References

1. David Remahl. [Polygenic traits result in a distribution that resembles a bell-shaped curve](#) . The creator of this work allows anyone to use it for any purpose including unrestricted redistribution, commercial use, and modification.

CONCEPT

21

Sex-linked Inheritance

- Define sex-linked trait.
- Explain the genetics of sex determination in humans.
- Explain sex-linked inheritance.

**Male or female?**

One of the exciting things about expecting a child is wondering if the baby will be a boy or a girl. There are many superstitions about how one might influence or predict the outcome. But what really determines if a baby is male or female? We now know that the gender of a baby is determined by a special pair of chromosomes known as the sex chromosomes.

Sex-linked Inheritance

What determines if a baby is a male or female? Recall that you have 23 pairs of chromosomes—and one of those pairs is the **sex chromosomes**. Everyone has two sex chromosomes. Your sex chromosomes can be X or Y. Females have two X chromosomes (XX), while males have one X chromosome and one Y chromosome (XY).

If a baby inherits an X chromosome from the father and an X chromosome from the mother, what will be the child's sex? The baby will have two X chromosomes, so it will be female. If the father's sperm carries the Y chromosome, the child will be male. Notice that a mother can only pass on an X chromosome, so the sex of the baby is determined by the father. The father has a 50 percent chance of passing on the Y or X chromosome, so there is a 50 percent chance that a child will be male, and there is a 50 percent chance a child will be female. This 50:50 chance occurs for each baby. A couple's first five children could all be boys. The sixth child still has a 50:50 chance of being a girl.

One special pattern of inheritance that doesn't fit Mendel's rules is **sex-linked inheritance**, referring to the inheritance of traits that are located on genes on the sex chromosomes. Since males and females do not have the same sex chromosomes, there will be differences between the sexes in how these **sex-linked traits**—traits linked to genes located on the sex chromosomes—are expressed. Sex-linked traits usually refer to traits due to genes on the X chromosome.

One example of a sex-linked trait is red-green colorblindness. People with this type of colorblindness cannot tell the difference between red and green. They often see these colors as shades of brown (**Figure 21.1**). Boys are much more likely to be colorblind than girls (**Table 21.1**). This is because colorblindness is a sex-linked, recessive trait.

Boys only have one X chromosome, so if that chromosome carries the gene for colorblindness, they will be colorblind. As girls have two X chromosomes, a girl can have one X chromosome with the colorblind gene and one X chromosome with a normal gene for color vision. Since colorblindness is recessive, the dominant normal gene will mask the recessive colorblind gene. Females with one colorblindness allele and one normal allele are referred to as **carriers**. They carry the allele but do not express it.

How would a female become colorblind? She would have to inherit two genes for colorblindness, which is very unlikely. Many sex-linked traits are inherited in a recessive manner.

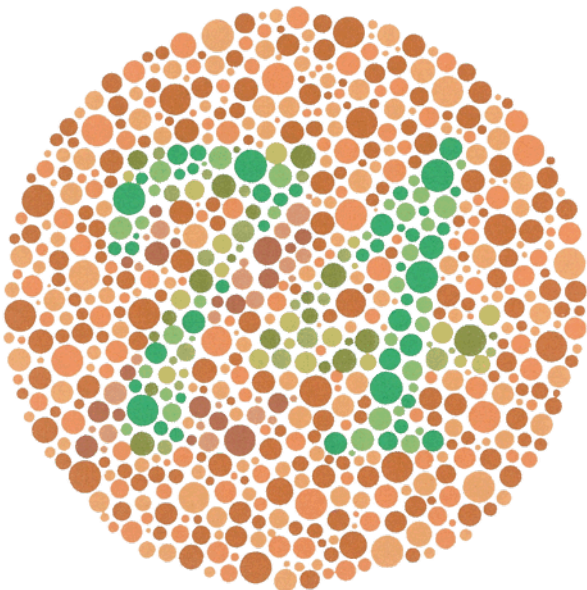


FIGURE 21.1

A person with red-green colorblindness would not be able to see the number.

TABLE 21.1: Cross Between a Female Carrier for Colorblindness and a Male with Normal Vision

	X^c	X
X	X^cX (carrier female)	XX (normal female)
Y	X^cY (colorblind male)	XY (normal male)

According to this Punnett square (**Table 21.1**), the son of a woman who carries the colorblindness trait and a male with normal vision has a 50% chance of being colorblind.

Summary

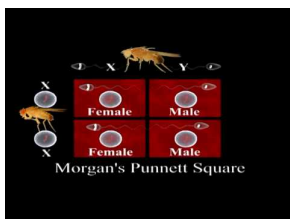
- Each individual has two sex chromosomes; females have two X chromosomes (XX), while males have one X chromosome and one Y chromosome (XY).
- Sex-linked traits are located on genes on the sex chromosomes.

Explore More

Use the resources below to answer the questions that follow.

Explore More I

- **Sex-linked Traits** at <http://www.youtube.com/watch?v=H1HaR47Dqfw> (5:16)



MEDIA

Click image to the left or use the URL below.

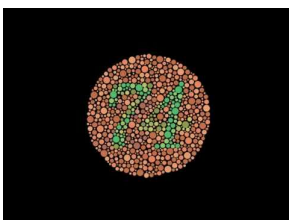
URL: <https://www.ck12.org/flx/render/embeddedobject/282>

1. What was unusual about the F_2 generations in Morgan's crosses?
2. According to Morgan, where is the gene for eye color located?
3. How did Morgan test his hypothesis on the location of the eye color gene?
4. What are three traits that humans have that are related to genes exclusive to the X-chromosome?

The "Morgan" referred to in the above clip is Thomas Hunt Morgan. You can find out more about him and his work here: <http://www.nature.com/scitable/topicpage/thomas-hunt-morgan-and-sex-linkage-452> .

Explore More II

- **Inheritance of Sex-linked Traits** at <http://www.youtube.com/watch?v=IJqFk-28G08> (4:49)



MEDIA

Click image to the left or use the URL below.

URL: <https://www.ck12.org/flx/render/embeddedobject/57305>

1. What are the three types of color blindness? How are they caused?
2. What is the "Law of Dominance"?
3. Can a woman have colorblindness if her father does not? Explain your answer fully.
4. A woman is color blind but her sister isn't. What does that tell you about their parents?

If you're still puzzled by sex-linked traits you can go to this site for more practice solving problems. Make sure you make good use of the "hints" on the site.

- **Sex-linked genes** at <http://www.ksu.edu/biology/pob/genetics/xlinked.htm>

Review

1. What are the sex chromosomes of a male and a female?
2. Explain why the father determines the sex of the child.
3. What is sex-linked inheritance?
4. A son cannot inherit colorblindness from his father. Why not?

References

1. . [A person with red-green colorblindness would not be able to see the number](#) . Public Domain

CONCEPT **22**

Genetic Disorders

- Describe the genetics of common human genetic disorders.



When is a cold not just a cold?

At some point in your life, you're bound to catch a cold. And there are ways to prevent catching a cold. But what if you couldn't prevent an illness? What if you were born with a disease? What if having a disease was actually due to your DNA? These are genetic diseases, and they can be very serious.

Human Genetic Disorders

Many **genetic disorders** are caused by mutations in one or a few genes. Others are caused by chromosomal mutations. Some human genetic disorders are X-linked or Y-linked, which means the faulty gene is carried on these sex chromosomes. Other genetic disorders are carried on one of the other 22 pairs of chromosomes; these chromosomes are known as **autosomes** or autosomal (non-sex) chromosomes. Some genetic disorders are due to new mutations, others can be inherited from your parents.

Autosomal Recessive Disorders

Some genetic disorders are caused by recessive alleles of a single gene on an autosome. An example of **autosomal recessive genetic disorders** are Tay-Sachs disease and cystic fibrosis. Children with **cystic fibrosis** have excessively thick mucus in their lungs, which makes it difficult for them to breathe. The inheritance of this recessive allele is the same as any other recessive allele, so a Punnett square can be used to predict the probability that two **carriers** of the disease will have a child with cystic fibrosis. Recall that carriers have the recessive allele for a trait but do not express the trait. What are the possible genotypes of the offspring in the following table (**Table 22.1**)? What are the possible phenotypes?

TABLE 22.1: Cross Between Two Carriers for Cystic Fibrosis

	F	f
F	FF (normal)	Ff (carrier)
f	Ff (carrier)	ff (affected)

According to this Punnett square, two parents that are carriers (Ff) of the cystic fibrosis gene have a 25% chance of having a child with cystic fibrosis (ff). The affected child must inherit two recessive alleles. The carrier parents are not affected.

Tay-Sachs disease is a severe genetic disorder in which affected children do not live to adulthood, so the gene is not passed from an affected individual. Carriers of the Tay-Sachs gene are not affected. How does a child become affected with Tay-Sachs?

Autosomal Dominant Disorders

Huntington's disease is an example of an **autosomal dominant disorder**. This means that if the dominant allele is present, then the person will express the disease. A child only has to inherit one dominant allele to have the disease.

The disease causes the brain's cells to break down, leading to muscle spasms and personality changes. Unlike most other genetic disorders, the symptoms usually do not become apparent until middle age. You can use a simple Punnett square to predict the inheritance of a dominant autosomal disorder, like Huntington's disease. If one parent has Huntington's disease, what is the chance of passing it on to the children? If you draw the Punnett square, you will find that there is a 50 percent chance of the disorder being passed on to the children.

Summary

- Autosomal recessive genetic disorders, such as cystic fibrosis, are caused by recessive alleles of a single gene on an autosome.
- Autosomal dominant genetic disorders, such as Huntington's disease, are caused by dominant alleles of a single gene on an autosome.

Explore More

Use the resource below to answer the questions that follow.

- **What are Genetic Disorders?** at <http://learn.genetics.utah.edu/content/disorders/>

1. What are multifactorial disorders? What is an example of a multifactorial disorder?

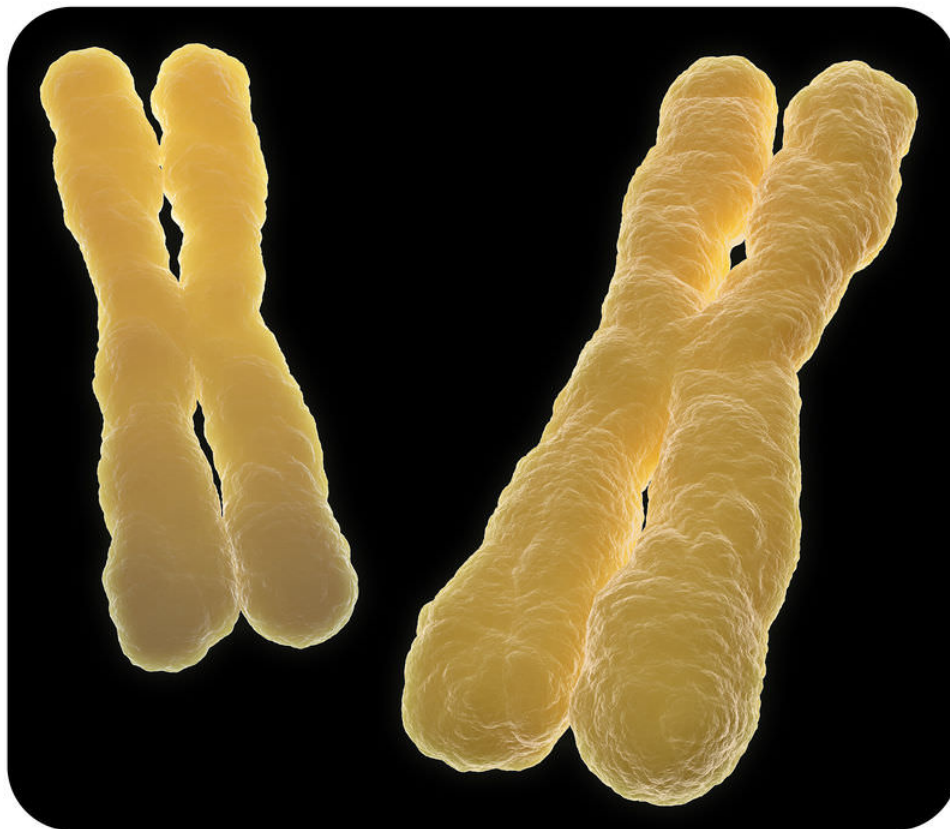
2. What are single-gene disorders? What is an example of a single-gene disorder?
3. What causes galactosemia? How is it diagnosed? How is it treated?
4. What causes Colon Cancer? What is a tumor suppressor gene?
5. What is newborn genetic screening? How is it carried out?

Review

1. Can you be a carrier of an autosomal recessive genetic disorder?
2. Can you be a carrier of an autosomal dominant genetic disorder?
3. One parent is a carrier of the cystic fibrosis gene, while the other parent does not carry the allele. Can their child have cystic fibrosis?

CONCEPT **23** Chromosomal Disorders

- Describe the genetics of Down syndrome.
- Explain how changes in chromosomes can cause disorders in humans.



Can you have too many chromosomes?

Yes, it's not a good thing to have extra chromosomes. An extra chromosome can be fatal to an embryo, in fact. In the case of a few chromosomes, however, a baby may be born with an extra chromosome. This child will have a chromosomal disorder.

Chromosomal Disorders

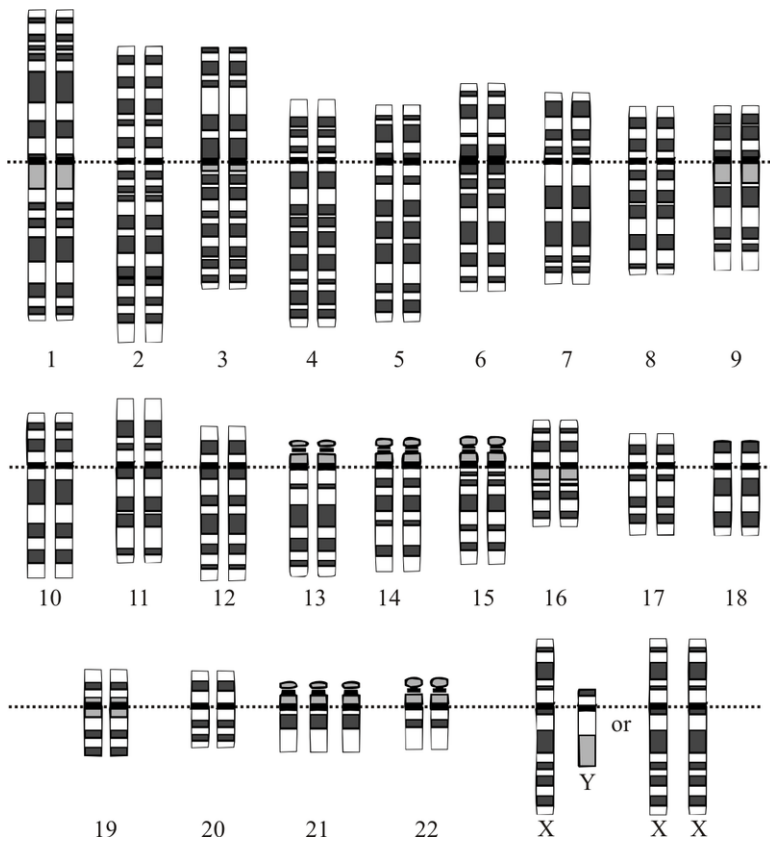
Some children are born with genetic defects that are not carried by a single gene. Instead, an error in a larger part of the chromosome or even in an entire chromosome causes the disorder. Usually the error happens when the egg or sperm is forming. Having extra chromosomes or damaged chromosomes can cause disorders.

Extra Chromosomes

One common example of an extra-chromosome disorder is **Down syndrome (Figure 23.1)**. Children with Down syndrome are mentally disabled and also have physical deformities. Down syndrome occurs when a baby receives an extra chromosome 21 from one of his or her parents. Usually, a child will receive one chromosome 21 from the mother and one chromosome 21 from the father. In an individual with Down syndrome, however, there are three


FIGURE 23.1

A child with Down syndrome.


FIGURE 23.2

Chromosomes of a person with Down Syndrome. Notice the extra chromosome 21.

copies of chromosome 21 (**Figure 23.2**). Therefore, Down syndrome is also known as Trisomy 21. These people have 47 total chromosomes.

Another example of a chromosomal disorder is **Klinefelter syndrome**, in which a male inherits an extra “X” chromosome. These individuals have an XXY genotype. They have underdeveloped sex organs and elongated limbs. They also have difficulty learning new things.

Outside of chromosome 21 and the sex chromosomes, most embryos with extra chromosomes do not usually survive. Because chromosomes carry many, many genes, a disruption of a chromosome can cause severe problems with the development of a fetus. Individuals with one (or more) fewer chromosome usually don't survive either. Can you explain why?

Damaged Chromosomes

Chromosomal disorders also occur when part of a chromosome becomes damaged. For example, if a tiny portion of chromosome 5 is missing, the individual will have *cri du chat* (cat's cry) syndrome. These individuals have misshapen facial features, and the infant's cry resembles a cat's cry.

Summary

- Changes in chromosome number can lead to disorders like Down syndrome.
- Chromosomal disorders also occur when part of a chromosome becomes damaged.

Explore More

Use the resources below to answer the questions that follow.

Explore More I

- **Down Syndrome** at <http://www.ygyh.org/ds/whatisit.htm>
1. Are all cases of Down Syndrome the result of inheritance?
 2. Do all cases of Down Syndrome have a complete extra chromosome? Explain your answer fully.
 3. How can a fetus be screened for Down syndrome?

Explore More II

- **Understanding Rare Chromosome Disorders** at <http://www.youtube.com/watch?v=k4Lps1kIyR0> (8:11)



MEDIA

Click image to the left or use the URL below.

URL: <https://www.ck12.org/flx/render/embeddedobject/57298>

1. What do all people diagnosed with a chromosome disorder share?
2. What is a clinical geneticist? What are they trained to do that is different from a regular doctor?
3. What is a karyotype?
4. Do chromosomal disorders always involve extra genetic material? Explain your answer.

Review

1. What is a chromosomal disorder?
2. Explain what causes Down Syndrome.
3. When do chromosomal defects occur?
4. What happens to most embryos with extra chromosomes? Explain your answer.

References

1. Image copyright Tomasz Markowski, 2014. [A child with Down syndrome](#) . Used under license from Shutterstock.com
2. Courtesy of the National Human Genome Research Institute. [Chromosomes of a person with Down Syndrome, with an extra chromosome21](#) . Public Domain

CONCEPT 24

Cloning

- Describe the process of animal cloning.
- Summarize the significance of Dolly.



Would you like to clone yourself?

Although it's illegal to clone humans in the United States, it is possible to clone many types of animals. What might be the consequences if we allowed human cloning?

Cloning

Cloning is the process of creating an exact genetic replica of an organism. The clone's DNA is exactly the same as the parent's DNA. Bacteria and other single-celled organisms have long been able to clone themselves through asexual reproduction. Plants can also reproduce asexually. In animals, however, cloning does not happen naturally. In 1997, that all changed when a sheep named Dolly was the first large mammal ever to be successfully cloned. Other animals can now also be cloned in a laboratory.

The process of producing an animal like Dolly starts with a single cell from the animal that is going to be cloned. Below are the steps involved in the process of cloning:

1. In the case of Dolly, cells from the mammary glands were taken from the adult that was to be cloned. But other somatic cells can be used. **Somatic cells** come from the body and are not gametes like sperm or egg.
2. The nucleus is removed from this cell.
3. The nucleus is placed in a donor egg that has had its nucleus removed. The nucleus must be removed from the donor egg to maintain the appropriate chromosome number.
4. The new cell is stimulated with an electric shock and embryo development begins, as if it were a normal **zygote**. The zygote is the first cell of a new organism.
5. The resulting embryo is implanted into a mother sheep, where it continues its development (**Figure 24.1**).

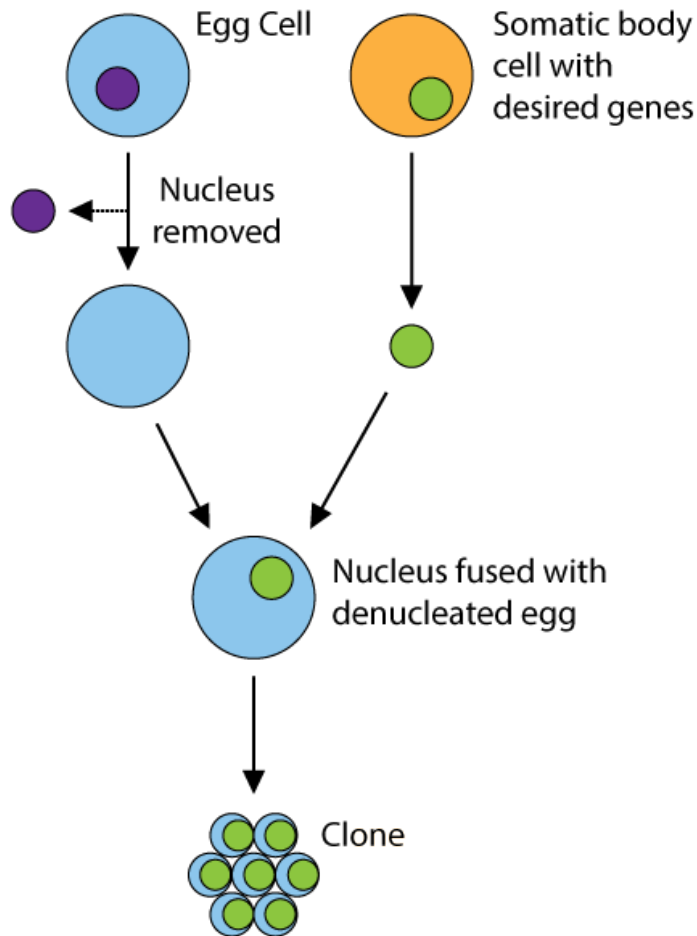


FIGURE 24.1

To clone an animal, a nucleus from the animal's cells are fused with an egg cell (from which the nucleus has been removed) from a donor, creating a new zygote.

Is Cloning Easy?

Cloning is not always successful. Most of the time, this cloning process does not result in a healthy adult animal. The process has to be repeated many times until it works. In fact, 277 tries were needed to produce Dolly. This high failure rate is one reason that human cloning is banned in the United States. In order to produce a cloned human, many attempts would result in the surrogate mothers experiencing miscarriages, stillbirths, or deformities in the infant. There are also many additional ethical considerations related to human cloning. Can you think of reasons why people are for or against cloning?

Summary

- Cloning, or creating an exact replica of an organism, is now possible for many animals.
- There are many ethical considerations related to human cloning, and it is now illegal to clone humans in the United States.

Explore More

Use the resource below to answer the questions that follow.

- **Click and Clone** at <http://learn.genetics.utah.edu/content/cloning/clickandclone/> .
1. What is the first step in cloning?
 2. How are the blunt and sharp pipettes used in the cloning process?
 3. How many cell divisions does the modified embryo go through before it is implanted in the surrogate mother?
 4. What step did scientists determine was crucial to the success of this process?

Review

1. Describe the process of creating an animal clone.
2. What are some reasons why human cloning is banned?

References

1. Zachary Wilson. [Process of how to clone an animal](#) .

CONCEPT

25

Human Genome Project

- Explain the significance of the Human Genome Project.



What is your genetic code?

The sequence of letters above represents bases in someone's DNA. It is now possible to find out a person's entire genetic code by determining all the bases in his or her DNA. What might be the benefits?

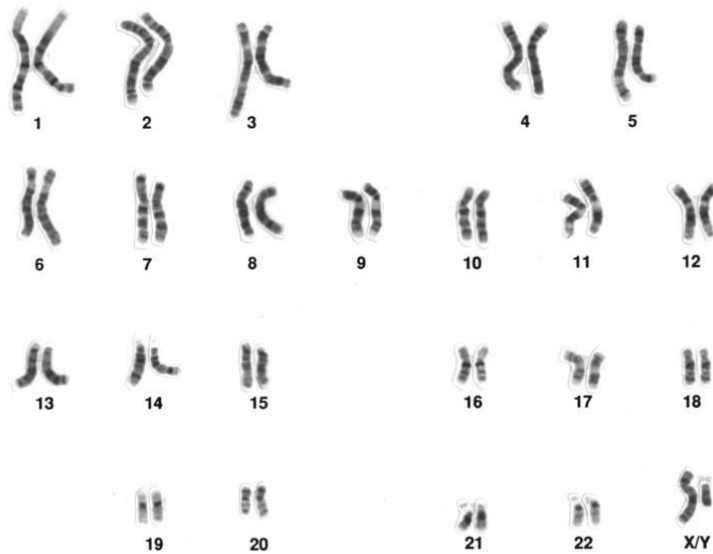
Human Genome Project

A person's **genome** is all of his or her genetic information. In other words, the human genome is all the information that makes us human. And unless you have an identical twin, your genome is unique. No one else has a genome just like yours, though all our genomes are similar.

The **Human Genome Project** (Figure 25.1) was an international effort to sequence all 3 billion bases that make up our DNA and to identify within this code more than 20,000 human genes. Scientists also completed a chromosome map, identifying where the genes are located on each of the chromosomes. The Human Genome Project was completed in 2003. Though the Human Genome Project is finished, analysis of the data will continue for many years. To say the Human Genome Project has been beneficial to mankind would be an understatement.

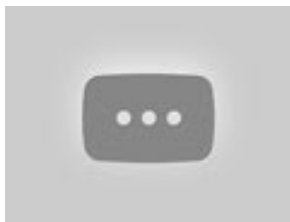
Exciting applications of the Human Genome Project include the following:

- The genetic basis for many diseases can be more easily determined. Now there are tests for over 1,000 genetic disorders.
- The technologies developed during this effort, and since the completion of this project, will reduce the cost of sequencing a person's genome. This may eventually allow many people to sequence their individual genome.
- Analysis of your own genome could determine if you are at risk for specific diseases.
- Knowing you might be genetically prone to a certain disease would allow you to make preventive lifestyle changes or have medical screenings.

**FIGURE 25.1**

To complete the Human Genome Project, all 23 pairs of chromosomes in the human body were sequenced. Each chromosome contains thousands of genes. This is a karyotype, a visual representation of an individual's chromosomes lined up by size.

The video *Our Molecular Selves* discusses the human genome, and is available at <http://www.genome.gov/25520211> or http://www.youtube.com/watch?v=_EK3g6px7Ik. *Genome, Unlocking Life's Code* is the Smithsonian National Museum of Natural History's exhibit on the human genome. See <http://unlockinglifescode.org> to visit the exhibit.

**MEDIA**

Click image to the left or use the URL below.

URL: <https://www.ck12.org/flx/render/embeddedobject/69173>

Summary

- The Human Genome Project involved an international effort to sequence all 3 billion bases that make up our DNA and to identify within this code more than 20,000 human genes.
- Analysis of your own genome could determine if you are at risk for specific diseases.

Explore More

Use the resources below to answer the questions that follow.

Explore More I

- **PCR Virtual Lab** at <http://learn.genetics.utah.edu/content/labs/pcr/>

1. How many copies of a DNA sequence can be made in a matter of hours using PCR?
2. About how much DNA do you need to start the PCR process?
3. Can you use the same primers for every DNA sequence you want to investigate? Why or why not?

4. How many "ingredients" go into your PCR tube? What are they?
5. What happens at 95°C during the PCR process?
6. What happens at 72°C during the PCR process?

Explore More II

- **Personal Genome Project** at <http://www.personalgenomes.org>

1. What is the goal of the personal genome project?
2. What information are they seeking besides a person's genome?
3. Why participate in the PGP?

Review

1. What is a person's genome?
2. Describe the Human Genome Project.
3. Would you want to know your own genome? Why or why not?

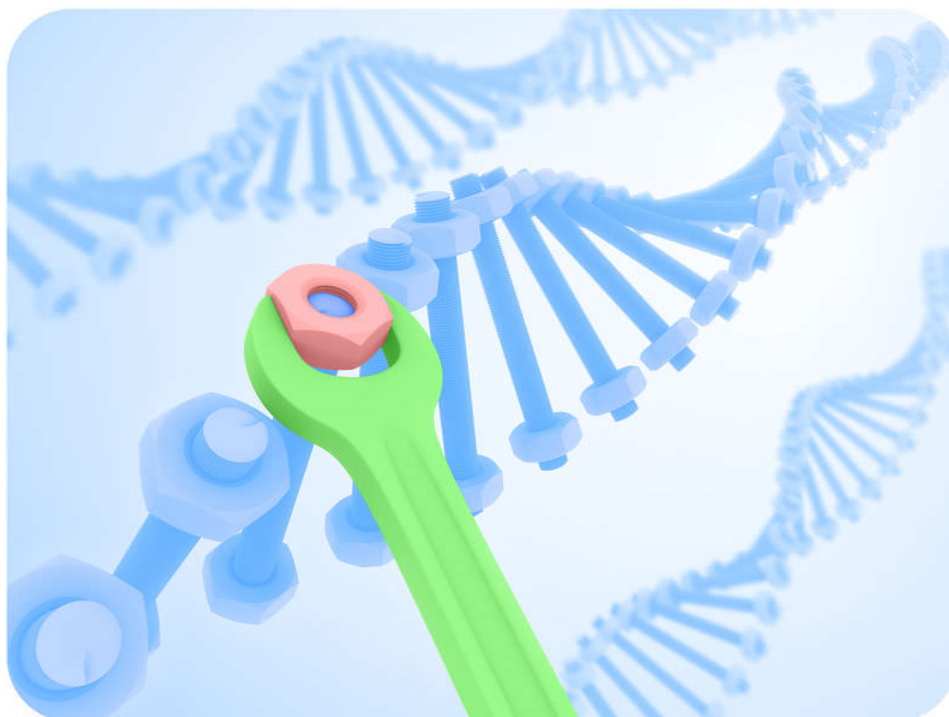
References

1. National Human Genome Research Institute. [Karyotype of human chromosomes](#) . Public Domain

CONCEPT 26

Gene Therapy

- Define gene therapy.
- Describe gene therapy.
- Distinguish *in vivo* gene therapy from *ex vivo* gene therapy.



Can doctors fix your DNA?

There are many genetic disorders that are due to a single gene. What if we could fix this faulty gene? With the development of gene therapy, that may eventually be possible for many types of genetic disorders.

Gene Therapy

Gene therapy is the insertion of genes into a person's cells to cure a genetic disorder. Could gene therapy be the cure for AIDS? No, AIDS is caused by a virus. Gene therapy only works to fix disorders caused by a faulty gene. The patient would have had this disorder from birth. Though gene therapy is still in experimental stages, the common use of this therapy may occur during your lifetime.

There are two main types of gene therapy:

1. One done inside the body (*in vivo*).
2. One done outside the body (*ex vivo*).

Both types of gene therapy use a **vector**, or carrier molecule for the gene. The vector helps incorporate the desired gene into the patient's DNA. Usually this vector is modified viral DNA in which the viral genes have been removed. Don't worry, the virus used in gene therapy has been deactivated.

In Vivo Gene Therapy

During *in vivo* gene therapy, done inside the body, the vector with the gene of interest is introduced directly into the patient and taken up by the patient's cells (**Figure 26.1**). For example, cystic fibrosis gene therapy is targeted at the respiratory system, so a solution with the vector can be sprayed into the patient's nose. Recently, *in vivo* gene therapy was also used to partially restore the vision of three young adults with a rare type of eye disease.

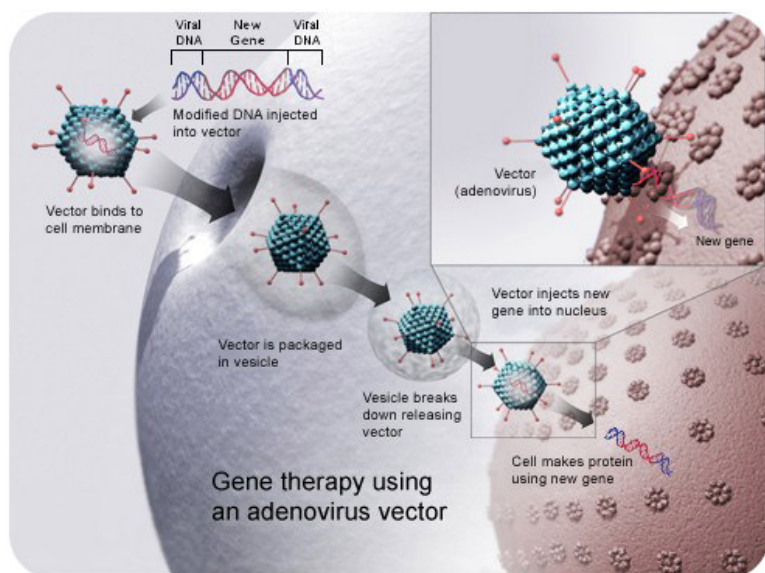


FIGURE 26.1

During gene therapy, adenovirus is a possible vector to carry the desired gene and insert it into the patient's DNA. A deactivated virus makes a useful vector for this purpose.

In ex vivo gene therapy, done outside the body, cells are removed from the patient and the proper gene is inserted using a virus as a vector. The modified cells are placed back into the patient.

One of the first uses of this type of gene therapy was in the treatment of a young girl with a rare genetic disease, adenosine deaminase deficiency, or ADA deficiency. People with this disorder are missing the ADA enzyme, which breaks down a toxin called deoxyadenosine. If the toxin is not broken down, it accumulates and destroys immune cells. As a result, individuals with ADA deficiency do not have a healthy immune system to fight off infections. In the gene therapy treatment for this disorder, bone marrow stem cells were taken from the girl's body, and the missing gene was inserted into these cells outside the body. Then the modified cells were put back into her bloodstream. This treatment successfully restored the function of her immune system, but only with repeated treatments.

Summary

- Gene therapy, the insertion of genes into a person's cells to cure a genetic disorder, can be *ex vivo* (outside the body) or *in vivo* (inside the body).
- Gene therapy is still in the experimental stages, but some trials have been successful.

Explore More

Use the resources below to answer the questions that follow.

- **What is Gene Therapy?** at <http://learn.genetics.utah.edu/content/genethrapy/gtintro/> .

- **Gene Delivery: Tools Of The Trade** at <http://learn.genetics.utah.edu/content/genetherapy/gttools/> .

1. What is an "ionic gradient"? How does cystic fibrosis affect the ionic gradient in cells? What effect does this have?
2. What five questions need to be answered to determine if a disease is a good candidate for gene therapy?
3. Why would an adenovirus vector be a bad choice for treating cystic fibrosis?
4. What is a good choice for a viral vector?

Review

1. What is gene therapy?
2. Could gene therapy someday cure the common cold? Why or why not?
3. What's the difference between *ex vivo* and *in vivo* gene therapy?

References

1. Courtesy of the National Institutes of Health. [Diagram of in vivo gene therapy using an adenovirus](#) .

CONCEPT **27**

Biotechnology in Agriculture

- Define transgenic crop.
- Explain how biotechnology can be used in agriculture.

**Have you ever eaten genetically engineered foods?**

Most likely, yes. The majority of the corn in the United States is genetically engineered. Corn syrup is used to sweeten many things, like this soft drink. Corn is also fed to the cows that provided this hamburger.

Biotechnology in Agriculture

Biotechnology is changing the genetic makeup of living things to make a useful product. Biotechnology has led scientists to develop useful applications in agriculture and food science. These include the development of **transgenic** crops. In transgenic crops, genes are placed into plants to give the crop a beneficial trait. Benefits include:

- Improved yield from crops.
- Increased resistance of crops to environmental stresses.
- Increased nutritional qualities of food crops.
- Improved taste, texture or appearance of food.
- Reduced dependence on fertilizers, insecticides, and other chemicals.

Crops are obviously dependent on environmental conditions. Drought can destroy crop yields, as can too much rain and floods. But what if crops could be developed to withstand these harsh conditions?

Biotechnology will allow the development of crops containing genes that will help them to withstand harsh conditions. For example, drought and salty soil are two significant factors affecting how well crops grow. But there are crops that can withstand these harsh conditions. Why? Probably because of that plant's genetics. So scientists are studying plants that can cope with these extreme conditions. They hope to identify and isolate the genes that control these beneficial traits. The genes could then be transferred into more desirable crops, with the hope of producing the same traits in those crops.

Thale cress (**Figure 27.1**), a species of *Arabidopsis* (*Arabidopsis thaliana*), is a tiny weed that has been extensively studied. It is often used for plant research because it is very easy to grow, and its DNA has been mapped. Scientists have identified a gene from this plant, At-DBF2, that gives the plant resistance to some environmental stresses. When this gene is inserted into tomato and tobacco cells, the cells were able to withstand environmental stresses like salt, drought, cold, and heat far better than ordinary cells. If these results prove successful in larger trials, then At-DBF2 genes could help in engineering crops that can better withstand harsh environments.



FIGURE 27.1

Thale cress (*Arabidopsis thaliana*).

Summary

- Genetic alteration can be used to change many different phenotypes of plants.

- Transgenic crops have extra genes that were placed into them to give the crop a beneficial trait.
- In the future, crops may be genetically altered to withstand harsh conditions.

Explore More

Use the resource below to answer the questions that follow.

- **How Do You Disable A Gene** at <http://www.youtube.com/watch?v=QEbVpj7EbwU> (6:19)



MEDIA

Click image to the left or use the URL below.

URL: <https://www.ck12.org/flx/render/embeddedobject/57299>

1. What approach do scientists use to disable genes in *Arabidopsis*? How does this work?
2. What do scientists use to insert DNA into *Arabidopsis*?
3. Can scientists insert whole genes into a plant's genome?
4. How are the *Arabidopsis* mutants valuable to botanists in general?

Review

1. What is a transgenic plant?
2. What are three examples of how biotechnology might be used in agriculture?

References

1. Quentin Groom. [Picture of thale cress, which is used for genetic research](#) . Public Domain