**4.1 Cell Division**

* Contrast cell division in prokaryotes and eukaryotes.

**Where do cells come from?**

No matter what the cell, all cells come from preexisting cells through the process of cell division. The cell may be the simplest bacterium or a complex muscle, bone, or blood cell. The cell may comprise the whole organism, or be just one cell of trillions.

### Cell Division

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

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

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

#### Cell Division in Prokaryotes

Most prokaryotic cells divide by the process of **binary fission**. A bacterial cell dividing this way is depicted in **Figure** [below](#x-ck12-QmlvLTA1LTAxLUJpbmFyeS1GaXNzaW9u) . You can also watch an animation of binary fission at this link: <http://en.wikipedia.org/wiki/File:Binary_fission_anim.gif>.

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

Binary fission can be described as a series of steps, although it is actually a continuous process. The steps are described below and also illustrated in **Figure** [below](#x-ck12-QmlvLTA1LTAyLVN0ZXBzLW9mLUJpbmFy) . They include DNA replication, chromosome segregation, and finally the separation into two daughter cells.

* Step 1: DNA Replication. Just before the cell divides, its DNA is copied in a process called DNA replication. This results in two identical chromosomes instead of just one. This step is necessary so that when the cell divides, each daughter cell will have its own chromosome.
* Step 2: Chromosome Segregation. The two chromosomes segregate, or separate, and move to opposite ends (known as "poles") of the cell. This occurs as each copy of DNA attaches to different parts of the cell membrane.
* Step 3: Separation. A new plasma membrane starts growing into the center of the cell, and the cytoplasm splits apart, forming two daughter cells. As the cell begins to pull apart, the new and the original chromosomes are separated. The two daughter cells that result are genetically identical to each other and to the parent cell. New cell wall must also form around the two cells.

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

#### Cell Division in Eukaryotes

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

1. The first step is **mitosis**, a multi-phase process in which the nucleus of the cell divides. During mitosis, the nuclear membrane breaks down and later reforms. The chromosomes are also sorted and separated to ensure that each daughter cell receives a diploid number (2 sets) of chromosomes. In humans, that number of chromosomes is 46 (23 pairs). Mitosis is described in greater detail in a subsequent concept.
2. The second major step is cytokinesis. As in prokaryotic cells, the cytoplasm must divide. Cytokinesis is the division of the cytoplasm in eukaryotic cells, resulting in two genetically identical daughter cells.

### Summary

* Cell division is part of the life cycle of virtually all cells. Cell division is the process in which one cell divides to form two new cells.
* Most prokaryotic cells divide by the process of binary fission.
* In eukaryotes, cell division occurs in two major steps: mitosis and cytokinesis.

### Practice I

* **Cell Division Quiz #1** at <http://www.neok12.com/quiz/CELDIV01>.

1. Describe the process of binary fission.
2. Compare the cells before and after the mitotic division.
3. What is cytokinesis?

# 4.2 Cell Cycle

* Identify the phases of the eukaryotic cell cycle.

**What is a cell's life like?**

The eukaryotic cell spends most of its "life" in interphase of the cell cycle, which can be subdivided into the three phases, G1, S and G2. During interphase, the cell does what it is supposed to do. Though cells have many common functions, such as DNA replication, they also have certain specific functions. That is, during the life of a heart cell, the cell would obviously perform certain different activities than a kidney cell or a liver cell.

### The Cell Cycle

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

#### The Eukaryotic Cell Cycle

The diagram in **Figure** [below](#x-ck12-QmlvLTA1LTAzLUV1a2FyeW90aWMtQ2Vs) represents the cell cycle of a eukaryotic cell. As you can see, the eukaryotic cell cycle has several phases. The **mitotic phase** (M) actually includes both mitosis and cytokinesis. This is when the nucleus and then the cytoplasm divide. The other three phases (G1, S, and G2) are generally grouped together as **interphase**. During interphase, the cell grows, performs routine life processes, and prepares to divide. These phases are discussed below. You can watch a eukaryotic cell going through these phases of the cell cycle at the following link: <http://www.cellsalive.com/cell_cycle.htm>.

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

#### Interphase

Interphase of the eukaryotic cell cycle can be subdivided into the following three phases, which are represented in **Figure** [above](#x-ck12-QmlvLTA1LTAzLUV1a2FyeW90aWMtQ2Vs) :

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

#### Control of the Cell Cycle

If the cell cycle occurred without regulation, cells might go from one phase to the next before they were ready. What controls the cell cycle? How does the cell know when to grow, synthesize DNA, and divide? The cell cycle is controlled mainly by regulatory proteins. These proteins control the cycle by signaling the cell to either start or delay the next phase of the cycle. They ensure that the cell completes the previous phase before moving on. Regulatory proteins control the cell cycle at key checkpoints, which are shown in **Figure** [below](#x-ck12-QmlvLTA1LTA0LVRoZS1DZWxsLUN5Y2xl) . There are a number of main checkpoints.

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

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

#### Cancer and the Cell Cycle

**Cancer** is a disease that occurs when the cell cycle is no longer regulated. This may happen because a cell’s DNA becomes damaged. Damage can occur due to exposure to hazards such as radiation or toxic chemicals. Cancerous cells generally divide much faster than normal cells. They may form a mass of abnormal cells called a **tumor** (see **Figure** [below](#x-ck12-QmlvLTA1LTA1LVR1bW9yLWNlbGxz) ). The rapidly dividing cells take up nutrients and space that normal cells need. This can damage tissues and organs and eventually lead to death.

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

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

Go to http://goo.gl/WzFCo2 for more content

### Summary

* The cell cycle is a repeating series of events that cells go through. It includes growth, DNA synthesis, and cell division. In eukaryotic cells, there are two growth phases, and cell division includes mitosis.
* The cell cycle is controlled by regulatory proteins at three key checkpoints in the cycle. The proteins signal the cell to either start or delay the next phase of the cycle.
* Cancer is a disease that occurs when the cell cycle is no longer regulated. Cancer cells grow rapidly and may form a mass of abnormal cells called a tumor.

# 4.3 Chromosomes

* Describe chromosomes and their role in mitosis.

**How is it assured that every cell in your body has the same DNA?**

Chromosomes, like those shown here, must form prior to cell division, to ensure that each daughter cell receives a complete set of genetic material. Essentially, each new cell receives half of each "X-shaped" chromosome.

### Chromosomes

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

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

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

Go to <http://goo.gl/2Zz0ua> for more content

#### Chromatids and the Centromere

DNA condenses and coils into the familiar X-shaped form of a chromosome, shown in **Figure** [below](#x-ck12-QmlvLTA1LTA2LUNocm9tb3NvbWU.) , only after it has replicated. (You can watch DNA coiling into a chromosome at the link below.) Because DNA has already replicated, each chromosome actually consists of two identical copies. The two copies are called sister **chromatids**. They are attached to one another at a region called the **centromere**. A remarkable animation can be viewed at <http://www.hhmi.org/biointeractive/media/DNAi_packaging_vo2-sm.mov>.

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

#### Chromosomes and Genes

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

#### Human Chromosomes

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

### Summary

* Chromosomes are coiled structures made of DNA and proteins.
* Chromosomes form after DNA replicates; prior to replication, DNA exists as chromatin.
* Chromosomes contain genes, which code for proteins.
* Human cells normally have 46 chromosomes, made up of two sets of chromosomes, one set inherited from each parent.

### Practice I

Use this resource to answer the questions that follow.

* **Chromosomes** at <http://johnkyrk.com/chromosomestructure.html>.

1. What is a chromosome?
2. What is chromatin?
3. What is a histone?

### Practice II

* **Quiz on Chromosomes** at <http://www.neok12.com/quiz/CELDIV04>.

# 4.4 Mitosis and Cytokinesis

* Outline the phases of mitosis.

**What is meant by the "division of the nucleus"?**

What do you think this colorful picture shows? If you guessed that it’s a picture of a cell undergoing cell division, you are right. But more specifically, the image is a lung cell stained with fluorescent dyes undergoing mitosis, during early anaphase.

### Mitosis and Cytokinesis

During **mitosis**, when the nucleus divides, the two chromatids that make up each chromosome separate from each other and move to opposite poles of the cell. This is shown in **Figure** [below](#x-ck12-QmlvLTA1LTA3LU1pdG9zaXMtaW4tRXVr) . You can watch an animation of the process at the following link: <http://www.biology.arizona.edu/Cell_bio/tutorials/cell_cycle/MitosisFlash.html>.

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

Mitosis actually occurs in four phases. The phases are called prophase, metaphase, anaphase, and telophase. They are shown in **Figure** [below](#x-ck12-QmlvLTA1LTA4LU1pdG9zaXMtZmxvd2No) and described in greater detail in the following sections.

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

#### Prophase

The first and longest phase of mitosis is **prophase**. During prophase, chromatin condenses into chromosomes, and the nuclear envelope, or membrane, breaks down. In animal cells, the **centrioles** near the nucleus begin to separate and move to opposite poles (sides) of the cell. As the centrioles move, a **spindle** starts to form between them. The spindle, shown in **Figure** [below](#x-ck12-QmlvLTA1LTA5LVNwaW5kbGUtYW5kLWNo) , consists of fibers made of microtubules.

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

#### Metaphase

During **metaphase**, spindle fibers attach to the centromere of each pair of sister chromatids (see **Figure** [below](#x-ck12-QmlvLTA1LTEwLU1ldGFwaGFzZS1EaWFn) ). The sister chromatids line up at the equator, or center, of the cell. The spindle fibers ensure that sister chromatids will separate and go to different daughter cells when the cell divides.

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

#### Anaphase

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

#### Telophase

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

#### Cytokinesis

Cytokinesis is the final stage of cell division in eukaryotes as well as prokaryotes. During cytokinesis, the cytoplasm splits in two and the cell divides. Cytokinesis occurs somewhat differently in plant and animal cells, as shown in **Figure** [below](#x-ck12-QmlvLTA1LTExLUN5dG9raW5lc2lz) . In animal cells, the plasma membrane of the parent cell pinches inward along the cell’s equator until two daughter cells form. In plant cells, a cell plate forms along the equator of the parent cell. Then, a new plasma membrane and cell wall form along each side of the cell plate.

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

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

Go to <http://goo.gl/7jMNhh> for more content

The four phases of mitosis. Can you describe what happens in each phase?

### Summary

* Cell division in eukaryotic cells includes mitosis, in which the nucleus divides, and cytokinesis, in which the cytoplasm divides and daughter cells form.
* Mitosis occurs in four phases, called prophase, metaphase, anaphase, and telophase.

### Practice I

* **Identify Mitosis Phases** at <http://www.neok12.com/quiz/CELDIV06>.
* **Animal Cell Mitosis** at <http://www.cellsalive.com/mitosis.htm>.

1. Why do cells undergo mitosis?
2. What are sister chromatids?
3. Define centromere and kinetochore.
4. What is the function of the mitotic spindle?
5. What are centrosomes and centrioles?
6. In what phase of mitosis do chromosomes line up at the center of the cell?
7. How are sister chromatids separated?
8. Compare cytokinesis in animal and plant cells.

# 4.5 Asexual vs. Sexual Reproduction

* Compare and contrast asexual and sexual reproduction.

**One parent or two?**

That is the main difference between sexual and asexual reproduction. Sexual reproduction just means combining genetic material from two parents. Asexual reproduction produces offspring genetically identical to the one parent.

### Reproduction: Asexual vs. Sexual

**Cell division** is how organisms grow and repair themselves. It is also how many organisms produce offspring. For many single-celled organisms, reproduction is a similar process. The parent cell simply divides to form two daughter cells that are identical to the parent. In many other organisms, two parents are involved, and the offspring are not identical to the parents. In fact, each offspring is unique. Look at the family in **Figure** [below](#x-ck12-QmlvLTA1LTEyLUZhbWlseQ..) . The children resemble their parents, but they are not identical to them. Instead, each has a unique combination of characteristics inherited from both parents.

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

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

#### Asexual Reproduction

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

* **Binary fission** occurs when a parent cell splits into two identical daughter cells of the same size.
* **Fragmentation** occurs when a parent organism breaks into fragments, or pieces, and each fragment develops into a new organism. Starfish, like the one in **Figure** [below](#x-ck12-QmlvLTA1LTAzLVN0YXJmaXNoLXllYXN0), reproduce this way. A new starfish can develop from a single ray, or arm. Starfish are also capable of sexual reproduction.
* **Budding** occurs when a parent cell forms a bubble-like bud. The bud stays attached to the parent cell while it grows and develops. When the bud is fully developed, it breaks away from the parent cell and forms a new organism. Budding in yeast is shown in **Figure** [beside](#x-ck12-QmlvLTA1LTAzLVN0YXJmaXNoLXllYXN0) .

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

#### Sexual Reproduction

**Sexual reproduction** involves two parents. As you can see from **Figure** [below](#x-ck12-QmlvLTA1LTE1LVNleHVhbC1SZXByb2R1) , in sexual reproduction, parents produce reproductive cells—called **gametes** —that unite to form an offspring. Gametes are **haploid** cells. This means they contain only half the number of chromosomes found in other cells of the organism. Gametes are produced by a type of cell division called **meiosis**, which is described in detail in a subsequent concept. The process in which two gametes unite is called **fertilization**. The fertilized cell that results is referred to as a **zygote**. A zygote is **diploid** cell, which means that it has twice the number of chromosomes as a gamete.

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

Go to <http://goo.gl/thshr3> for more content

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

### Summary

* Asexual reproduction involves one parent and produces offspring that are genetically identical to each other and to the parent.
* Sexual reproduction involves two parents and produces offspring that are genetically unique.
* During sexual reproduction, two haploid gametes join in the process of fertilization to produce a diploid zygote.
* Meiosis is the type of cell division that produces gametes.

### Practice

1. What type of organisms can benefit from asexual reproduction?
2. Give two examples of organisms that reproduce by binary fission.
3. What is the difference between budding and fragmentation?
4. What is sexual reproduction?
5. How many chromosomes are in a human gamete?

# 4.6 Meiosis

* Give an overview of sexual reproduction, and outline the phases of meiosis.

**How do you make a cell with half the DNA?**

Meiosis. This allows cells to have half the number of chromosomes, so two of these cells can come back together to form a new organism with the complete number of chromosomes. This process not only helps produce gametes, it also ensures genetic variation.

### Meiosis

The process that produces haploid gametes is meiosis. **Meiosis** is a type of cell division in which the number of chromosomes is reduced by half. It occurs only in certain special cells of the organisms. During meiosis, homologous chromosomes separate, and **haploid** cells form that have only one chromosome from each pair. Two cell divisions occur during meiosis, and a total of four haploid cells are produced. The two cell divisions are called meiosis I and meiosis II. The overall process of meiosis is summarized in **Figure** [below](#x-ck12-QmlvLTA1LTE2LU92ZXJ2aWV3LW9mLW1l) . You can watch an animation of meiosis at this link: [http://www.youtube.com/watch?v=D1\_-mQS\_FZ0&#38;feature=related](http://www.youtube.com/watch?v=D1_-mQS_FZ0&feature=related) .

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

#### Phases of Meiosis

Meiosis I begins after DNA replicates during interphase of the cell cycle. In both meiosis I and meiosis II, cells go through the same four phases as mitosis - prophase, metaphase, anaphase and telophase. However, there are important differences between meiosis I and mitosis. The flowchart in **Figure** [below](#x-ck12-QmlvLTA1LTE3LXBoYXNlcy1vZi1tZWlv) shows what happens in both meiosis I and II. You can follow the changes in the flowchart as you read about them below.

Compare meiosis I in this flowchart with the figure from the "Cell Division IV: Mitosis and Cytokinesis" concept. How does meiosis I differ from mitosis? Notice at the beginning of meiosis (prophase I), homologous chromosomes exchange segments of DNA. This is known as **crossing-over**, and is unique to this phase of meiosis.

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

Go to <http://goo.gl/K0FOpo> for more content

#### Meiosis I

1. Prophase I: **Homologous chromosomes** pair up and cross-over, which is unique to prophase I. In prophase of mitosis and meiosis II, homologous chromosomes do not form pairs in this way.
2. Metaphase I: The paired chromosomes line up along the equator (middle) of the cell. In metaphase of mitosis and meiosis II, it is sister chromatids that line up along the equator of the cell.
3. Anaphase I: Spindle fibers shorten, and the chromosomes of each homologous pair start to separate from each other. One chromosome of each pair moves toward one pole of the cell.
4. Telophase I and Cytokinesis: The spindle breaks down, and new nuclear membranes form. The cytoplasm of the cell divides, and two unique haploid daughter cells result. Both daughter cells go on to meiosis II. The DNA does not replicate between meiosis I and meiosis II.

#### Meiosis II

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

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

Go to <http://goo.gl/UZOEYB> for more content

You can watch an animation of meiosis at this link: [http://www.youtube.com/watch?v=D1\_-mQS\_FZ0&#38;feature=related](http://www.youtube.com/watch?v=D1_-mQS_FZ0&feature=related) .

### Summary

* Meiosis is the type of cell division that produces gametes.
* Meiosis involves two cell divisions and produces four haploid cells.
* Sexual reproduction has the potential to produce tremendous genetic variation in offspring. This is due in part to crossing-over during meiosis.

### Practice I

Use this resource to answer the questions that follow.

* **Meiosis** at <http://www.concord.org/activities/meiosis>.
* **Meiosis** at <http://www.neok12.com/diagram/Cell-Division-03.htm>.
* **Meiosis** at <http://johnkyrk.com/meiosis.html>.
* **Identify Meiosis phases** at <http://www.neok12.com/quiz/CELDIV08>.
* **Quiz on Meiosis** at <http://www.neok12.com/quiz/CELDIV07>.

1. What is a fundamental goal of meiosis?
2. What does 2n refer to?
3. Describe synapsis, crossing-over, and chiasmata.
4. Describe the main difference between metaphase I and metaphase II. ( Hint : DNA alignment)
5. List four differences between meiosis and mitosis.

# 4.7 Gametogenesis

* Explain why sexual reproduction leads to variation in offspring.

**What's the biggest cell on Earth?**

The ostrich egg - unfertilized, of course. Yes, this egg, just like a human ovum, is just one cell. The egg shell membrane encloses the nucleus containing the genetic material and the cytoplasm.

### Gametogenesis

At the end of meiosis, four haploid cells have been produced, but the cells are not yet gametes. The cells need to develop before they become mature gametes capable of fertilization. The development of haploid cells into gametes is called **gametogenesis**.

How much DNA is in a gamete? The sperm cell forms by meiosis and spermatogenesis. Because it forms by meiosis, the sperm cell has only half as much DNA as a body cell. Notice the three distinct segments: a head piece, a flagella tail and a midpiece of mostly mitochondria. What is the role of each section?

Gametogenesis may differ between males and females. Male gametes are called **sperm**. Female gametes are called **eggs**. In human males, for example, the process that produces mature sperm cells is called **spermatogenesis**. During this process, sperm cells grow a tail and gain the ability to “swim,” like the human sperm cell shown in **Figure** [below](#x-ck12-QmlvLTA1LTE4LXNwZXJtLWFuZC1lZ2c.) . In human females, the process that produces mature eggs is called **oogenesis**. Just one egg is produced from the four haploid cells that result from meiosis. The single egg is a very large cell, as you can see from the human egg in **Figure** [below](#x-ck12-QmlvLTA1LTE4LXNwZXJtLWFuZC1lZ2c.) .

A human sperm is a tiny cell with a tail. A human egg is much larger. Both cells are mature haploid gametes that are capable of fertilization. What process is shown in this photograph? Notice the sperm with the head piece containing the genetic material, a flagella tail that propels the sperm, and a midpiece of mostly mitochondria, supplying ATP.

#### Spermatogenesis and Oogenesis

During spermatogenesis, primary **spermatocytes** go through the first cell division of meiosis to produce secondary spermatocytes. These are haploid cells. Secondary spermatocytes then quickly complete the meiotic division to become **spermatids**, which are also haploid cells. The four haploid cells produced from meiosis develop a flagellum tail and compact head piece to become mature sperm cells, capable of swimming and fertilizing an egg. The compact head, which has lost most of its cytoplasm, is key in the formation of a streamlined shape. The middle piece of the sperm, connecting the head to the tail, contains many mitochondria, providing energy to the cell. The sperm cell essentially contributes only DNA to the zygote.

On the other hand, the egg provides the other half of the DNA, but also organelles, building blocks for compounds such as proteins and nucleic acids, and other necessary materials. The egg, being much larger than a sperm cell, contains almost all of the cytoplasm a developing embryo will have during its first few days of life. Therefore, oogenesis is a much more complicated process than spermatogenesis.

Oogenesis begins before birth and is not completed until after fertilization. Oogenesis begins when **oogonia** (singular, oogonium), which are the immature eggs that form in the ovaries before birth and have the diploid number of chromosomes, undergo mitosis to form primary **oocytes**, also with the diploid number. Oogenesis proceeds as a primary oocyte undergoes the first cell division of meiosis to form secondary oocytes with the haploid number of chromosomes. A secondary oocyte only undergoes the second meiotic cell division to form a haploid ovum if it is fertilized by a sperm. The one egg cell that results from meiosis contains most of the cytoplasm, nutrients, and organelles. This unequal distribution of materials produces one large cell, and one cell with little more than DNA. This other cell, known as a **polar body**, eventually breaks down. The larger cell undergoes meiosis II, once again producing a large cell and a polar body. The large cell develops into the mature gamete, called an **ovum** ( **Figure** [below](#x-ck12-T09nZW5lc2lzMQ..) ).

Maturation of the ovum. Notice only 1 mature ovum, or egg, forms during meiosis from the primary oocyte. Three polar bodies may form during oogenesis. These polar bodies will not form mature gametes. Conversely, four haploid spermatids form during meiosis from the primary spermatocyte.

### Summary

* Meiosis is a step during spermatogenesis and oogenesis.
* Spermatogenesis produces four haploid sperm cells, while oogenesis produces one mature ovum.

### Practice

Use this resource to answer the questions that follow.

* <http://www.hippocampus.org/Biology>Biology for AP\* Search: **Gamete Production.**

1. Describe briefly how gametes are produced. What is a germ cell?
2. Distinguish between a spermatogonium and an oogonium.
3. Distinguish between a spermatocyte and a spermatid.
4. What is a follicle?
5. Describe the corpus luteum.
6. Compare and contrast oogenesis and spermatogenesis.

# 4.8 Genetic Variation

* Explain why sexual reproduction leads to variation in offspring.

**What helps ensure the survival of a species?**

Genetic variation. It is this variation that is the essence of evolution. Without genetic differences among individuals, "survival of the fittest" would not be likely. Either all survive, or all perish.

### Genetic Variation

**Sexual reproduction** results in infinite possibilities of genetic variation. In other words, sexual reproduction results in offspring that are genetically unique. They differ from both parents and also from each other. This occurs for a number of reasons.

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

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

#### Crossing-Over

As mentioned above, crossing-over occurs during prophase I, and it is the exchange of genetic material between non-sister chromatids of homologous chromosomes. Recall during prophase I, homologous chromosomes line up in pairs, gene-for-gene down their entire length, forming a configuration with four chromatids, known as a **tetrad**. At this point, the chromatids are very close to each other and some material from two chromatids switch chromosomes, that is, the material breaks off and reattaches at the same position on the homologous chromosome ( **Figure** [below](#x-ck12-QmlvMDYtMDItMDQ.) ). This exchange of genetic material can happen many times within the same pair of homologous chromosomes, creating unique combinations of genes. This process is also known as **recombination**.

Crossing-over. A maternal strand of DNA is shown in red. Paternal strand of DNA is shown in blue. Crossing over produces two chromosomes that have not previously existed. The process of recombination involves the breakage and rejoining of parental chromosomes (M, F). This results in the generation of novel chromosomes (C1, C2) that share DNA from both parents.

#### Independent Assortment and Random Fertilization

In humans, there are over 8 million configurations in which the chromosomes can line up during metaphase I of meiosis. It is the specific processes of meiosis, resulting in four unique haploid cells, that result in these many combinations. This independent assortment, in which the chromosome inherited from either the father or mother can sort into any gamete, produces the potential for tremendous genetic variation. Together with random fertilization, more possibilities for genetic variation exist between any two people than the number of individuals alive today. Sexual reproduction is the random fertilization of a gamete from the female using a gamete from the male. In humans, over 8 million (2 23 ) chromosome combinations exist in the production of gametes in both the male and female. A sperm cell, with over 8 million chromosome combinations, fertilizes an egg cell, which also has over 8 million chromosome combinations. That is over 64 trillion unique combinations, not counting the unique combinations produced by crossing-over. In other words, each human couple could produce a child with over 64 trillion unique chromosome combinations!

See How Cells Divide: Mitosis vs. Meiosis ( <http://www.pbs.org/wgbh/nova/miracle/divide.html>) for an animation comparing the two processes.

### Summary

* Sexual reproduction has the potential to produce tremendous genetic variation in offspring.
* This variation is due to independent assortment and crossing-over during meiosis, and random union of gametes during fertilization.

### Practice

1. List the sources of genetic variation.
2. What are the advantages of sexual reproduction?

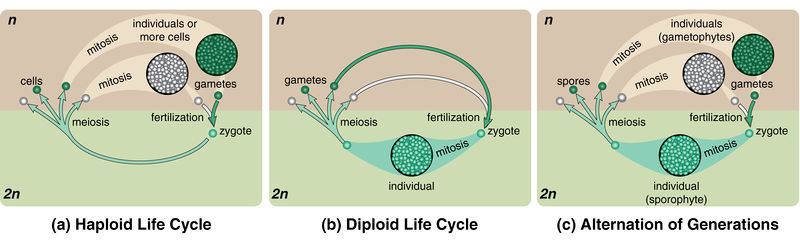
# 4.9 Reproductive Life Cycles

* Define life cycle, and identify different types of sexual life cycles.

**Young to old. A life cycle?**

Not in the biological sense. Life cycles describe the amount of DNA present at a specific stage or time in the life of an organism. Is there a haploid or diploid amount of DNA? That is the key question.

### Life Cycles

Sexual reproduction occurs in a cycle. Diploid parents produce haploid gametes that unite and develop into diploid adults, which repeat the cycle. This series of life stages and events that a sexually reproducing organism goes through is called its **life cycle**. Sexually reproducing organisms can have different types of life cycles. Three are represented in **Figure** [below](#x-ck12-QmlvSS0wNS0xOS1zZXh1YWwtbGlmZS1j) and described following sections.

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

#### Haploid Life Cycle

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

#### Diploid Life Cycle

Organisms with a **diploid life cycle** spend the majority of their lives as diploid adults. When they are ready to reproduce, they undergo meiosis and produce haploid gametes. Gametes then unite in fertilization and form a diploid zygote, which immediately enters G 1 of the cell cycle. Next, the zygote's DNA is replicated. Finally, the processes of mitosis and cytokinesis produce two genetically identical diploid cells. Through repeated rounds of growth and division, this organism becomes a diploid adult and the cycle continues. Can you think of an organism with a diploid life cycle? ( Hint: What type of life cycle do humans have?)

#### Alternation of Generations

Plants, algae, and some protists have a life cycle that alternates between diploid and haploid phases, known as **alternation of generations**. In plants, the life cycle alternates between the diploid sporophyte and haploid gametophyte. Spore forming cells in the diploid sporophyte undergo meiosis to produce **spores**, a haploid reproductive cell. Spores can develop into an adult without fusing with another cell. The spores give rise to a multicellular haploid **gametophyte**, which produce gametes by mitosis. The gametes fuse, producing a diploid zygote, which grow into the diploid sporophyte. These life cycles may be quite complicated. You can read about them in additional concepts.

### Summary

* A life cycle is the sequence of stages an organisms goes through from one generation to the next. Organisms that reproduce sexually can have different types of life cycles, such as haploid or diploid life cycles.

### Practice

1. What are the "hallmarks" of the sexual life cycle?
2. Why might some animals switch between sexual and asexual reproduction?

# 4.10 Mendel's Pea Plants

* Explain why and how Mendel studied pea plants.

**What's so interesting about pea plants?**

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

### Mendel and His Pea Plants

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

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

Go to <http://goo.gl/0tmY8O> for more content.

Gregor Mendel was born in 1822 and grew up on his parents’ farm in Austria. He did well in school and became a monk. He also went to the University of Vienna, where he studied science and math. His professors encouraged him to learn science through experimentation and to use math to make sense of his results. Mendel is best known for his experiments with the pea plant Pisum sativum (see **Figure** [below](#x-ck12-QmlvLTA2LTAxLUdyZWdvci1NZW5kZWw.) ). You can watch a video about Mendel and his research at the following link: <http://www.metacafe.com/watch/hl-19246625/milestones_in_science_engineering_gregor_mendel_and_classical_genetics/>.

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

#### Blending Theory of Inheritance

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

#### Why Study Pea Plants?

Why did Mendel choose common, garden-variety pea plants for his experiments? Pea plants are a good choice because they are fast growing and easy to raise. They also have several visible characteristics that may vary. These characteristics, which are shown in **Figure** [beside](#x-ck12-QmlvLTA2LTAyLU1lbmRlbC1TZXZlbi1D), include seed form and color, flower color, pod form and color, placement of pods and flowers on stems, and stem length. Each characteristic has two common values. For example, seed form may be round or wrinkled, and flower color may be white or purple (violet).

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

#### Controlling Pollination

To research how characteristics are passed from parents to offspring, Mendel needed to control pollination. **Pollination** is the fertilization step in the sexual reproduction of plants. **Pollen** consists of tiny grains that are the male gametes of plants. They are produced by a male flower part called the **anther** (see **Figure** [below](#x-ck12-QmlvLTA2LTAzLVBhcnRzLW9mLWEtZmxv) ). Pollination occurs when pollen is transferred from the anther to the stigma of the same or another flower. The **stigma** is a female part of a flower. It passes the pollen grains to female gametes in the ovary.

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

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

### Summary

* Gregor Mendel experimented with pea plants to learn how characteristics are passed from parents to offspring.
* Mendel’s discoveries formed the basis of genetics, the science of heredity.
* Cross-pollination produces hybrids.

### Making Connections

Go to <http://goo.gl/cTz3yW> for more multimedia content.

### Practice

1. Why did Mendel choose to work with pea plants?
2. What were the pea plant traits Mendel studied?
3. What are the stamen and carpel?
4. How did Mendel cross-pollinate plants?

# 4.11 Mendel's First Set of Experiments

* Describe the results of Mendel’s experiments.

**Peas. Some round and some wrinkled. Why?**

That's what Mendel asked. He noticed peas were always round or wrinkled, but never anything else. Seed shape was one of the traits Mendel studied in his first set of experiments.

### Mendel’s First Set of Experiments

Mendel first experimented with just one characteristic of a pea plant at a time. He began with flower color. As shown in **Figure** [below](#x-ck12-QmlvLTA2LTA0LUNoYXJ0LVNob3dpbmct), Mendel cross-pollinated purple- and white-flowered parent plants. The parent plants in the experiments are referred to as the **P** (for parent) **generation**. You can explore an interactive animation of Mendel’s first set of experiments at this link: <http://www2.edc.org/weblabs/Mendel/mendel.html>.

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

#### F1 and F2 Generations

The offspring of the P generation are called the **F1** (for filial, or “offspring”) **generation**. As you can see from **Figure** [above](#x-ck12-QmlvLTA2LTA0LUNoYXJ0LVNob3dpbmct), all of the plants in the F1 generation had purple flowers. None of them had white flowers. Mendel wondered what had happened to the white-flower characteristic. He assumed some type of inherited factor produces white flowers and some other inherited factor produces purple flowers. Did the white-flower factor just disappear in the F1 generation? If so, then the offspring of the F1 generation—called the **F2 generation** —should all have purple flowers like their parents. To test this prediction, Mendel allowed the F1 generation plants to self-pollinate. He was surprised by the results. Some of the F2 generation plants had white flowers. He studied hundreds of F2 generation plants, and for every three purple-flowered plants, there was an average of one white-flowered plant.

#### Law of Segregation

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

### Summary

* Mendel first researched one characteristic at a time. This led to his law of segregation. This law states that each characteristic is controlled by two factors, which separate and go to different gametes when an organism reproduces.

### Making Connections

Go to <http://goo.gl/Gzhoac> for more multimedia content.

### Practice I

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

1. Define a true-breeding strain. How did Mendel make sure the plants were true-breeding?
2. What is a monohybrid cross?
3. Define allele. Give an example.
4. What is the difference between homozygous and heterozygous?
5. Why did Mendel not observe any white flowered plants in the F 1 generation of his experiment?
6. Why was Mendel able to observe white flowered plants in the F 2 generation of his experiment?
7. How many alleles of a gene are in a gamete?
8. Explain Mendel's Law of Segregation.

### Practice II

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

# 4.12 Mendel's Second Set of Experiments

* Describe the results of Mendel’s experiments.

**Round and green, round and yellow, wrinkled and green, or wrinkled and yellow?**

Can two traits be inherited together? Or are all traits inherited separately? Mendel asked these questions after his first round of experiments.

### Mendel’s Second Set of Experiments

After observing the results of his first set of experiments, Mendel wondered whether different characteristics are inherited together. For example, are purple flowers and tall stems always inherited together? Or do these two characteristics show up in different combinations in offspring? To answer these questions, Mendel next investigated two characteristics at a time. For example, he crossed plants with yellow round seeds and plants with green wrinkled seeds. The results of this cross, which is a **dihybrid cross**, are shown in **Figure** [below](#x-ck12-QmlvLTA2LTA1LURpaHlicmlkLUNyb3Nz) .

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

#### F1 and F2 Generations

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

#### Law of Independent Assortment

Mendel repeated this experiment with other combinations of characteristics, such as flower color and stem length. Each time, the results were the same as those in **Figure** [above](#x-ck12-QmlvLTA2LTA1LURpaHlicmlkLUNyb3Nz). The results of Mendel’s second set of experiments led to his second law. This is the **law of independent assortment**. It states that factors controlling different characteristics are inherited independently of each other.

### Summary

* After his first set of experiments, Mendel researched two characteristics at a time. This led to his law of independent assortment. This law states that the factors controlling different characteristics are inherited independently of each other.

### Making Connections

Go to <http://goo.gl/V1Qo0b> for more multimedia content.

### Practice I

Use this resource to answer the questions that follow.

* **The Geniverse Lab** at <http://www.concord.org/activities/geniverse-lab>.

1. What is a dihybrid cross? Give an example.
2. What would a YYRR plant look like?
3. When did Mendel observe a 9:3:3:1 ratio in the F 2 generation?
4. What does Mendel's second law state?

# 4.13 Mendel's Laws and Genetics

* State Mendel’s laws of segregation and independent assortment.

**Do you look like your parents?**

You probably have some characteristics or traits in common with each of your parents. Mendel's work provided the basis to understand the passing of traits from one generation to the next.

### Mendel’s Laws and Genetics

You might think that Mendel’s discoveries would have made a big impact on science as soon as he made them. But you would be wrong. Why? Mendel never published his work. Charles Darwin published his landmark book on evolution in 1859, not long after Mendel had discovered his laws, but Darwin knew nothing of Mendel’s discoveries. As a result, Darwin didn’t understand heredity. This made his arguments about evolution less convincing to many people. This example shows why it is important for scientists to communicate the results of their investigations.

#### Rediscovering Mendel’s Work

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

#### Genetics of Inheritance

Today, we known that characteristics of organisms are controlled by genes on chromosomes (see **Figure** [below](#x-ck12-QmlvLTA2LTA2LVBhcnRzLW9mLWEtY2hy) ). The position of a gene on a chromosome is called its **locus**. In sexually reproducing organisms, each individual has two copies of the same gene, as there are two versions of the same chromosome ( **homologous chromosomes** ). One copy comes from each parent. The gene for a characteristic may have different versions, but the different versions are always at the same locus. The different versions are called **alleles**. For example, in pea plants, there is a purple-flower allele ( B ) and a white-flower allele ( b ). Different alleles account for much of the variation in the characteristics of organisms.

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

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

#### Genotype and Phenotype

When gametes unite during fertilization, the resulting zygote inherits two alleles for each gene. One allele comes from each parent. The alleles an individual inherits make up the individual’s **genotype**. The two alleles may be the same or different. As shown in **Table** [below](#x-ck12-dGFibGU6Zmxvd2VyLWNvbG9yLXBlYXM.) , an organism with two alleles of the same type ( BB or bb ) is called a **homozygote**. An organism with two different alleles ( Bb ) is called a **heterozygote**. This results in three possible genotypes.

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

The expression of an organism’s genotype produces its **phenotype**. The phenotype refers to the organism’s characteristics, such as purple or white flowers. As you can see from **Table** [above](#x-ck12-dGFibGU6Zmxvd2VyLWNvbG9yLXBlYXM.) , different genotypes may produce the same phenotype. For example, BB and Bb genotypes both produce plants with purple flowers. Why does this happen? In a Bb heterozygote, only the B allele is expressed, so the b allele doesn’t influence the phenotype. In general, when only one of two alleles is expressed in the phenotype, the expressed allele is called the **dominant** allele. The allele that isn’t expressed is called the **recessive** allele.

#### How Mendel Worked Backward to Get Ahead

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

### Summary

* Mendel’s work was rediscovered in 1900. Soon after that, genes and alleles were discovered. This allowed Mendel’s laws to be stated in terms of the inheritance of alleles.
* The gene for a characteristic may have different versions. These different versions of a gene are known as alleles.
* Alleles for different genes assort independently during meiosis.
* The alleles an individual inherits make up the individual’s genotype. The individual may be homozygous (two of the same alleles) or heterozygous (two different alleles).
* The expression of an organism’s genotype produces its phenotype.
* When only one of two alleles is expressed, the expressed allele is the dominant allele, and the allele that isn’t expressed is the recessive allele.
* Mendel used the percentage of phenotypes in offspring to understand how characteristics are inherited.

### Making Connections

Go to <http://goo.gl/vt64ku> for more content

### Practice I

* **Modern Genetics** at <http://www.concord.org/activities/modern-genetics>.

1. What is an allele?
2. Define genotype and phenotype.
3. When is a person heterozygous or homozygous?

# 4.14 Probability and Inheritance

* Explain how probability is related to inheritance.

**What are the odds of landing on 7 again?**

Not as high as inheriting an allele from a parent. Probability plays a big role in determining the chance of inheriting an allele from a parent. It is similar to tossing a coin. What's the chance of the coin landing on heads?

### Probability

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

**Probability** is the likelihood, or chance, that a certain event will occur. The easiest way to understand probability is with coin tosses (see **Figure** [below](#x-ck12-QmlvLTA2LTA3LUNvaW4tZmxpcA..) ). When you toss a coin, the chance of a head turning up is 50 percent. This is because a coin has only two sides, so there is an equal chance of a head or tail turning up on any given toss.

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

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

#### Probability and Inheritance

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

#### Probability and Gamete Formation

How is gamete formation like tossing a coin? Consider Mendel’s purple-flowered pea plants again. Assume that a plant is heterozygous for the flower-color allele, so it has the genotype Bb (see **Figure** [below](#x-ck12-QmlvLTA2LTA4LUdhbWV0ZS1mb3JtYXRp) ). During meiosis, homologous chromosomes, and the alleles they carry, segregate and go to different gametes. Therefore, when the Bb pea plant forms gametes, the B and b alleles segregate and go to different gametes. As a result, half the gametes produced by the Bb parent will have the B allele and half will have the b allele. Based on the rules of probability, any given gamete of this parent has a 50 percent chance of having the B allele and a 50 percent chance of having the b allele.

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

#### Probability and Fertilization

Which of these gametes joins in fertilization with the gamete of another parent plant? This is a matter of chance, like tossing a coin. Thus, we can assume that either type of gamete—one with the B allele or one with the b allele—has an equal chance of uniting with any of the gametes produced by the other parent. Now assume that the other parent is also Bb . If gametes of two Bb parents unite, what is the chance of the offspring having one of each allele like the parents ( Bb )? What is the chance of them having a different combination of alleles than the parents (either BB or bb )? To answer these questions, geneticists use a simple tool called a Punnett square, which is the focus of the next concept.

### Summary

* Probability is the chance that a certain event will occur. For example, the probability of a head turning up on any given coin toss is 50 percent.
* Probability can be used to predict the chance of gametes and offspring having certain alleles.

### Making Connections

Go to <http://goo.gl/w7nDlG> for more content

### Practice

Use this resource to answer the questions that follow.

* **Fundamentals of Inheritance** at <http://www.biologie.uni-hamburg.de/b-online/library/falk/Inherit/Inherit.htm>.

1. Define probability as a sentence.
2. Define probability as a fraction.
3. What is the probability of cutting a deck of playing cards and getting an ace?
4. How can you determine the probability of two independent events that occur together?
5. What is the probability that two heterozygous individuals will have offspring with attached earlobes?

# 4.15 Punnett Squares

* Describe how to use a Punnett square.

**What do you get when you cross an apple and an orange?**

Though the above fruit may not result, it would be nice to scientifically predict what would result. Predicting the possible genotypes and phenotypes from a genetic cross is often aided by a Punnett square.

### Punnett Squares

A **Punnett square** is a chart that allows you to easily determine the expected percentage of different genotypes in the offspring of two parents. An example of a Punnett square for pea plants is shown in **Figure** [below](#x-ck12-QmlvLTA2LTA5LVB1cnBsZS13aGl0ZS1m) . In this example, both parents are **heterozygous** for flower color ( Bb ). The **gametes** produced by the male parent are at the top of the chart, and the gametes produced by the female parent are along the side. The different possible combinations of **alleles** in their offspring are determined by filling in the cells of the Punnett square with the correct letters (alleles). At the link below, you can watch an animation in which Reginald Punnett, inventor of the Punnett square, explains the purpose of his invention and how to use it. <http://www.dnalc.org/view/16192-Animation-5-Genetic-inheritance-follows-rules-.html>

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

Go to <http://goo.gl/FHbz17> for more content

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

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

#### Predicting Offspring Genotypes

In the cross shown in **Figure** [above](#x-ck12-QmlvLTA2LTA5LVB1cnBsZS13aGl0ZS1m) , you can see that one out of four offspring (25 percent) has the **genotype** BB , one out of four (25 percent) has the genotype bb , and two out of four (50 percent) have the genotype Bb . These percentages of genotypes are what you would expect in any cross between two heterozygous parents. Of course, when just four offspring are produced, the actual percentages of genotypes may vary by chance from the expected percentages. However, if you considered hundreds of such crosses and thousands of offspring, you would get very close to the expected results, just like tossing a coin.

#### Predicting Offspring Phenotypes

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

#### Determining Missing Genotypes

A Punnett square can also be used to determine a missing genotype based on the other genotypes involved in a cross. Suppose you have a parent plant with purple flowers and a parent plant with white flowers. Because the b allele is recessive, you know that the white-flowered parent must have the genotype bb . The purple-flowered parent, on the other hand, could have either the BB or the Bb genotype. The Punnett square in **Figure** [below](#x-ck12-QmlvLTA2LTEwLXdoaXRlLWFuZC1wdXJw) shows this cross. The question marks (?) in the chart could be either B or b alleles.

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

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

#### Punnett Square for Two Characteristics

When you consider more than one characteristic at a time, using a Punnett square is more complicated. This is because many more combinations of alleles are possible. For example, with two genes each having two alleles, an individual has four alleles, and these four alleles can occur in 16 different combinations. This is illustrated for pea plants in **Figure** [below](#x-ck12-QmlvLTA2LTExLUNyb3BwZWQtcHVubmV0) . In this cross, known as a **dihybrid cross** , both parents are heterozygous for pod color ( Gg ) and seed color ( Yy ).

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

### Summary

* A Punnett square is a chart that allows you to determine the expected percentages of different genotypes in the offspring of two parents.
* A Punnett square allows the prediction of the percentages of phenotypes in the offspring of a cross from known genotypes.
* A Punnett square can be used to determine a missing genotype based on the other genotypes involved in a cross.

### Making Connections

Go to <http://goo.gl/87aFSE> for more content

### Practice I

Review <http://goo.gl/IHmIBl> and answer the questions below.

1. What is a Punnett square?
2. What is the size of a Punnett square used in a dihybrid cross?
3. Define the following terms: alleles, genotype, phenotype, genome.

# 4.16 Non-Mendelian Inheritance

* Describe complex patterns of inheritance.

**Green, blue, brown, black, hazel, violet, or grey. What color are your eyes?**

Of course human eyes do not come in multi-color, but they do come in many colors. How do eyes come in so many colors? That brings us to complex inheritance patterns, known as non-Mendelian inheritance. Many times inheritance is more complicated than the simple patterns observed by Mendel.

### Non-Mendelian Inheritance

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

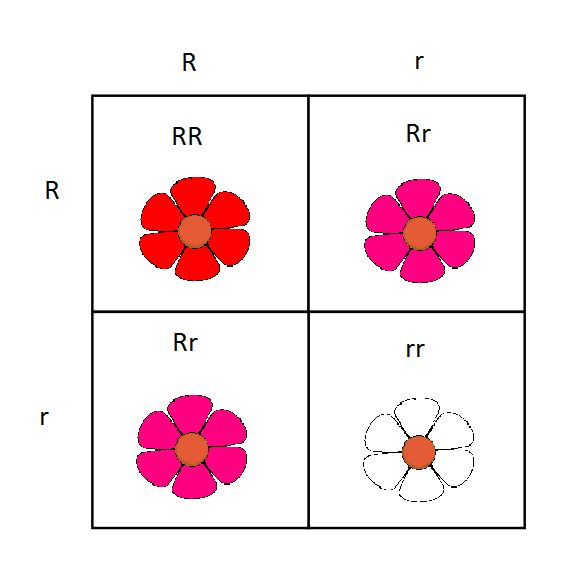
A characteristic may be controlled by one gene with two alleles, but the two alleles may have a different relationship than the simple dominant-recessive relationship that you have read about so far. For example, the two alleles may have a codominant or incompletely dominant relationship. The former is illustrated by the flower in **Figure** [below](#x-ck12-QmlvLTA2LTEyLVJlZC1hbmQtd2hpdGUt) , and the latter in **Figure** [below](#x-ck12-QmlvNy0yLTc.) .

#### Codominance

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

Codominance. The flower has red and white petals because the red and white alleles are codominant.

#### Incomplete Dominance

**Incomplete dominance** occurs when the phenotype of the offspring is somewhere in between the phenotypes of both parents; a completely dominant allele does not occur. For example, when red snapdragons (C R C R ) are crossed with white snapdragons (C W C W ), the F 1 hybrids are all pink hetrozygotes for flower color (C R C W ). The pink color is an intermediate between the two parent colors. When two F 1 (C R C W ) hybrids are crossed they will produce red, pink, and white flowers. The genotype of an organism with incomplete dominance can be determined from its phenotype ( **Figure** [below](#x-ck12-QmlvNy0yLTc.) ).

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

#### Multiple Alleles

Many genes have multiple (more than two) alleles. An example is **ABO blood type** in humans. There are three common alleles for the gene that controls this characteristic. The alleles I A and I B are dominant over i. A person who is homozygous recessive ii has type O blood. Homozygous dominant I A I A or heterozygous dominant I A i have type A blood, and homozygous dominant I B I B or heterozygous dominant I B i have type B blood. I A I B people have type AB blood, because the A and B alleles are codominant. Type A and type B parents can have a type AB child. Type A and type B parents can also have a child with Type O blood, if they are both heterozygous ( I B i , I A i ).

* Type A blood: I A I A , I A i
* Type B blood: I B I B , I B i
* Type AB blood: I A I B
* Type O blood: ii

#### Polygenic Characteristics

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

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

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

#### Effects of Environment on Phenotype

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

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

### Summary

* Many characteristics have more complex inheritance patterns than those studied by Mendel. They are complicated by factors such as codominance, incomplete dominance, multiple alleles, and environmental influences.

### Practice

1. What is the genotype of a pink carnation?
2. What are the alleles for blood type in humans?
3. How is skin color in humans determined?
4. Define pleiotrophy.

# 4.17 Human Genome

* Define the human genome.

**All these ACGTs. What are they?**

Over three billion of them from a human form the human genome - the human genetic material - all the information needed to encode a human being. It would take about 9.5 years to read out loud - without stopping - the more than three billion pairs of bases in one person's genome.

### The Human Genome

What makes each one of us unique? You could argue that the environment plays a role, and it does to some extent. But most would agree that your parents have something to do with your uniqueness. In fact, it is our **genes** that make each one of us unique – or at least genetically unique. We all have the genes that make us human: the genes for skin and bones, eyes and ears, fingers and toes, and so on. However, we all have different skin colors, different bone sizes, different eye colors and different ear shapes. In fact, even though we have the same genes, the products of these genes work a little differently in most of us. And that is what makes us unique.

The **human genome** is the **genome** - all the DNA - of Homo sapiens. Humans have about 3 billion bases of information, divided into roughly 20,000 to 22,000 genes, which are spread among non-coding sequences and distributed among 24 distinct chromosomes (22 **autosomes** plus the X and Y **sex chromosomes** ) ( [below](#x-ck12-QmlvLTA4LTAxLWh1bWFuLWdlbm9tZQ..) ). The genome is all of the hereditary information encoded in the DNA, including the genes and non-coding sequences.

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

Thanks to the **Human Genome Project**, scientists now know the DNA sequence of the entire human genome. The Human Genome Project is an international project that includes scientists from around the world. It began in 1990, and by 2003, scientists had sequenced all 3 billion base pairs of human DNA. Now they are trying to identify all the genes in the sequence. The Human Genome Project has produced a reference sequence of the human genome. The human genome consists of protein-coding **exons**, associated **introns** and regulatory sequences, genes that encode other RNA molecules, and “junk” DNA - regions in which no function as yet been identified.

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

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

Go to <http://goo.gl/4oG4GD> for more content

#### ENCODE: The Encyclopedia of DNA Elements

In September 2012, ENCODE, The **Enc**yclopedia **o**f **D**NA **E**lements, was announced. ENCODE was a colossal project, involving over 440 scientists in 32 labs the world-over, whose goal was to understand the human genome. It had been thought that about 80% of the human genome was "junk" DNA. ENCODE has established that this is not true. Now it is thought that about 80% of the genome is active. In fact, much of the human genome is regulatory sequences, on/off switches that tell our genes what to do and when to do it. Dr. Eric Green, director of the National Human Genome Research Institute of the National Institutes of Health which organized this project, states, "It's this incredible choreography going on, of a modest number of genes and an immense number of ... switches that are choreographing how those genes are used."

It is now thought that at least three-quarters of the genome is involved in making RNA, and most of this RNA appears to help regulate gene activity. Scientists have also identified about 4 million sites where proteins bind to DNA and act in a regulatory capacity. These new findings demonstrate that the human genome has remarkable and precise, and complex, controls over the expression of genetic information within a cell.

See ENCODE data describes function of human genome at <http://www.genome.gov/27549810>for additional information.

### Summary

* The human genome consists of about 3 billion base pairs of DNA.
* In 2003, the Human Genome Project finished sequencing all 3 billion base pairs.

### Practice

Use this resource to answer the questions that follow.

* <http://www.hippocampus.org/Biology>Non-Majors Biology Search: **Human Genome Project**

1. What were 3 goals of the Human Genome Project?
2. How many genes are on chromosome #1?
3. How big is the human genome?
4. How much genetic variation is there among people?
5. How much of the genome is not part of any gene?

# 4.18 Human Chromosomes and Genes

* Describe human chromosomes and genes.

**Coiled bundles of DNA and proteins, containing hundreds or thousands of genes. What are these things?**

Chromosomes. These ensure that each cell receives the proper amount of DNA during cell division. And usually people have 46 of them, 23 from each parent.

### Chromosomes and Genes

Each species has a characteristic number of chromosomes. **Chromosomes** are coiled structures made of DNA and proteins called **histones** ( **Figure** [below](#x-ck12-QmlvOS0xLTI.) ). Chromosomes are the form of the genetic material of a cell during cell division. See the "Chromosomes" section for additional information.

The human genome has 23 pairs of chromosomes located in the nucleus of somatic cells. Each chromosome is composed of genes and other DNA wound around histones (proteins) into a tightly coiled molecule.

The human species is characterized by 23 pairs of chromosomes, as shown in **Figure** [below](#x-ck12-QmlvLTA4LTAzLWh1bWFuLWNocm9tb3Nv) . You can watch a short animation about human chromosomes at this link: <http://www.dnalc.org/view/15520-DNA-is-organized-into-46-chromosomes-including-sex-chromosomes-3D-animation.html>.

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

#### Autosomes

Of the 23 pairs of human chromosomes, 22 pairs are autosomes (numbers 1–22 in **Figure** [above](#x-ck12-QmlvLTA4LTAzLWh1bWFuLWNocm9tb3Nv) ). **Autosomes** are chromosomes that contain genes for characteristics that are unrelated to sex. These chromosomes are the same in males and females. The great majority of human genes are located on autosomes. At the link below, you can click on any human chromosome to see which traits its genes control. <http://www.ornl.gov/sci/techresources/Human_Genome/posters/chromosome/chooser.shtml>

#### Sex Chromosomes

The remaining pair of human chromosomes consists of the **sex chromosomes** , X and Y. Females have two X chromosomes, and males have one X and one Y chromosome. In females, one of the X chromosomes in each cell is inactivated and known as a Barr body. This ensures that females, like males, have only one functioning copy of the X chromosome in each cell. As you can see from **Figure** [above](#x-ck12-QmlvLTA4LTAyLXNleC1jaHJvbW9zb21l) and **Figure** [above](#x-ck12-QmlvLTA4LTAzLWh1bWFuLWNocm9tb3Nv) , the X chromosome is much larger than the Y chromosome. The X chromosome has about 2,000 genes, whereas the Y chromosome has fewer than 100, none of which are essential to survival. Virtually all of the X chromosome genes are unrelated to sex. Only the Y chromosome contains genes that determine sex. A single Y chromosome gene, called **SRY** (which stands for sex-determining region Y gene), triggers an embryo to develop into a male. Without a Y chromosome, an individual develops into a female, so you can think of female as the default sex of the human species. Can you think of a reason why the Y chromosome is so much smaller than the X chromosome? At the link that follows, you can watch an animation that explains why: <http://www.hhmi.org/biointeractive/gender/Y_evolution.html>.

#### Human Genes

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

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

### Summary

* Humans have 23 pairs of chromosomes. Of these, 22 pairs are autosomes.
* The X and Y chromosomes are the sex chromosomes. Females have two X chromosomes, and males have one X and one Y.
* Human chromosomes contain a total of 20,000 to 22,000 genes, the majority of which have two or more alleles.

### Practice

Use these resources to answer the questions that follow.

* <http://www.hippocampus.org/Biology>Biology for AP\* Search: **The Chromosome Theory**

1. What is the chromosome theory of inheritance?
2. Distinguish between an autosome and a sex chromosome.

* <http://www.hippocampus.org/Biology>Biology for AP\* Search: **Sex Chromosomes**

1. What determines the sex of a baby?
2. Define an X-linked gene.
3. Why is it more likely that a male will display an X-linked trait than a female?

# 4.19 Genetic Linkage

* Explain linkage and linkage maps.

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

For a pair of hands, the above image may suggest a certain type of linkage. For genes, it might suggest that they are very hard to separate.

### Linkage

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

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

#### Sex-Linked Genes

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

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

Go to <http://goo.gl/EsHRXM> for more content

#### Mapping Linkage

Linkage can be assessed by determining how often crossing-over occurs between two genes on the same chromosome. Genes on different (nonhomologous) chromosomes are not linked. They assort independently during meiosis, so they have a 50 percent chance of ending up in different gametes. If genes show up in different gametes less than 50 percent of the time (that is, they tend to be inherited together), they are assumed to be on the same (homologous) chromosome. They may be separated by crossing-over, but this is likely to occur less than 50 percent of the time. The lower the frequency of crossing-over, the closer together on the same chromosome the genes are presumed to be. Frequencies of crossing-over can be used to construct a linkage map like the one in **Figure** [below](#x-ck12-QmlvLTA4LTA0LUxpbmthZ2UtbWFw) . A **linkage map** shows the locations of genes on a chromosome.

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

### Summary

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

### Practice II

Use this resource to answer the questions that follow.

* **T. H. Morgan** at <http://www.dnalc.org/resources/nobel/morgan.html>.

1. Who was Thomas Hunt Morgan?
2. Why must each chromosome contain many genes?
3. What are linked genes?
4. Describe the the relationship between cross-over events and linked genes.
5. Define recombination.
6. Why did Mendel not observe cross-over offspring?
7. What is a linkage map?

# 4.20 Mendelian Inheritance in Humans

* Describe inheritance in humans for autosomal and X-linked traits.

**What number can you see?**

Red-green colorblindness is a common inherited trait in humans. About 1 in 10 men have some form of color blindness, however, very few women are color blind. Why?

### Mendelian Inheritance in Humans

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

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

#### Autosomal Traits

Autosomal traits are controlled by genes on one of the 22 human autosomes. Consider earlobe attachment. A single autosomal gene with two alleles determines whether you have attached earlobes or free-hanging earlobes. The allele for free-hanging earlobes ( F ) is dominant to the allele for attached earlobes ( f ). Other single-gene autosomal traits include widow’s peak and hitchhiker’s thumb. The dominant and recessive forms of these traits are shown in **Figure** [below](#x-ck12-QmlvLTA4LTA1LUVhcmxvYmUtYXR0YWNo) . Which form of these traits do you have? What are your possible genotypes for the traits? The chart in **Figure** [below](#x-ck12-QmlvLTA4LTA1LUVhcmxvYmUtYXR0YWNo) is called a **pedigree**. It shows how the earlobe trait was passed from generation to generation within a family. Pedigrees are useful tools for studying inheritance patterns.

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

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

Other single-gene autosomal traits include widow's peak and hitchhiker's thumb. The dominant and recessive forms of these traits are shown in **Figure** [below](#x-ck12-QmlvLTA4LTA2LUF1dG9zb21hbC10cmFp) . Which form of these traits do you have? What are your possible genotypes for the traits?

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

#### Sex-Linked Traits

Traits controlled by genes on the sex chromosomes are called **sex-linked traits**, or **X-linked traits** in the case of the X chromosome. Single-gene X-linked traits have a different pattern of inheritance than single-gene autosomal traits. Do you know why? It’s because males have just one X chromosome. In addition, they always inherit their X chromosome from their mother, and they pass it on to all their daughters but none of their sons. This is illustrated in **Figure** [below](#x-ck12-QmlvLTA4LTA3LVNleC1DZWxscw..) .

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

Because males have just one X chromosome, they have only one allele for any X-linked trait. Therefore, a recessive X-linked allele is always expressed in males. Because females have two X chromosomes, they have two alleles for any X-linked trait. Therefore, they must inherit two copies of the recessive allele to express the recessive trait. This explains why X-linked recessive traits are less common in females than males. An example of a recessive X-linked trait is **red-green color blindness**. People with this trait cannot distinguish between the colors red and green. More than one recessive gene on the X chromosome codes for this trait, which is fairly common in males but relatively rare in females ( **Figure** [below](#x-ck12-QmlvLTA4LTA4LWNvbG9yYmxpbmRuZXNz) ). At the following link, you can watch an animation about another X-linked recessive trait called hemophilia A: <http://www.dnalc.org/view/16315-Animation-13-Mendelian-laws-apply-to-human-beings-.html>.

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

### Summary

* A minority of human traits are controlled by single genes with two alleles.
* They have different inheritance patterns depending on whether they are controlled by autosomal or X-linked genes.

### Practice I

Use <http://goo.gl/REM7bj> to answer the questions.

1. What is an X-linked gene? Give an example.
2. Will a color blind man always pass the color blind allele to daughters? Why or why not?
3. How can the son of a color blind man have color blindness?
4. What is meant by a female "carrier"?
5. A homozygous freckled man marries a non-freckled woman. If freckles are dominant, will their children have freckles? Explain your answer.
6. Using F and f , what are the genotypes of the parents? What are the genotypes of their gametes?

# 4.21 Genetic Disorders

* Describe genetic disorders caused by mutations or abnormal numbers of chromosomes.

**Is being short-statured inherited?**

It can be. Achondroplasia is the most common form of dwarfism in humans, and it is caused by a dominant mutation. The mutation can be passed from one generation to the next.

### Genetic Disorders

Many **genetic disorders** are caused by mutations in one or a few genes. Other genetic disorders are caused by abnormal numbers of chromosomes.

#### Genetic Disorders Caused by Mutations

The **Table** [below](#x-ck12-dGFibGU6Z2VuZXRpYy1kaXNvcmRlcnM.) lists several genetic disorders caused by mutations in just one gene. Some of the disorders are caused by mutations in autosomal genes, others by mutations in X-linked genes. Which disorder would you expect to be more common in males than females? You can watch a video about genetic disorders caused by mutations at this link: <http://www.pbs.org/wgbh/nova/programs/ht/rv/2809_03.html>.

You can click on any human chromosome at this link to see the genetic disorders associated with it: <http://www.ornl.gov/sci/techresources/Human_Genome/posters/chromosome/chooser.shtml>.

| **Genetic Disorder** | **Direct Effect of Mutation** | **Signs and Symptoms of the Disorder** | **Mode of Inheritance** |
| --- | --- | --- | --- |
| Marfan syndrome | defective protein in connective tissue | heart and bone defects and unusually long, slender limbs and fingers | autosomal dominant |
| Sickle cell anemia | abnormal hemoglobin protein in red blood cells | sickle-shaped red blood cells that clog tiny blood vessels, causing pain and damaging organs and joints | autosomal recessive |
| Vitamin D-resistant rickets | lack of a substance needed for bones to absorb minerals | soft bones that easily become deformed, leading to bowed legs and other skeletal deformities | X-linked dominant |
| Hemophilia A | reduced activity of a protein needed for blood clotting | internal and external bleeding that occurs easily and is difficult to control | X-linked recessive |

Few genetic disorders are controlled by dominant alleles. A mutant dominant allele is expressed in every individual who inherits even one copy of it. If it causes a serious disorder, affected people may die young and fail to reproduce. Therefore, the mutant dominant allele is likely to die out of the population. A mutant recessive allele, such as the allele that causes sickle cell anemia (see **Figure** [below](#x-ck12-QmlvLTA4LTExLXJlZC1ibG9vZC1jZWxs) and the link that follows), is not expressed in people who inherit just one copy of it. These people are called **carriers**. They do not have the disorder themselves, but they carry the mutant allele and can pass it to their offspring. Thus, the allele is likely to pass on to the next generation rather than die out. <http://www.dnalc.org/resources/3d/17-sickle-cell.html>

Sickle-Shaped and Normal Red Blood Cells. Sickle cell anemia is an autosomal recessive disorder. The mutation that causes the disorder affects just one amino acid in a single protein, but it has serious consequences for the affected person. This photo shows the sickle shape of red blood cells in people with sickle cell anemia.

Cystic Fibrosis and Tay-Sachs disease are two additional severe genetic disorders. They are discussed in the following video: <http://www.youtube.com/watch?v=8s4he3wLgkM&feature=related>(9:31). Tay-Sachs is further discussed at <http://www.youtube.com/watch?v=1RO0LOgHbIo&feature=channel>(3:13) and <http://www.youtube.com/watch?v=6zNj5LdDuTA>(2:01).

#### Chromosomal Disorders

Mistakes may occur during meiosis that result in **nondisjunction**. This is the failure of replicated chromosomes to separate during meiosis (the animation at the link below shows how this happens). Some of the resulting gametes will be missing a chromosome, while others will have an extra copy of the chromosome. If such gametes are fertilized and form zygotes, they usually do not survive. If they do survive, the individuals are likely to have serious genetic disorders. **Table** [below](#x-ck12-dGFibGU6YWJub3JtYWwtY2hyb21vc29t) lists several genetic disorders that are caused by abnormal numbers of chromosomes. Most chromosomal disorders involve the X chromosome. Look back at the X and Y chromosomes and you will see why. The X and Y chromosomes are very different in size, so nondisjunction of the sex chromosomes occurs relatively often. <http://learn.genetics.utah.edu/content/begin/traits/predictdisorder/index.html>

| **Genetic Disorder** | **Genotype** | **Phenotypic Effects** |
| --- | --- | --- |
| Down syndrome | extra copy (complete or partial) of chromosome 21 (see **Figure** [below](#x-ck12-Qy1CaW8tMDMtMTEtRG93bi1zeW5kcm9t) ) | developmental delays, distinctive facial appearance, and other abnormalities (see **Figure** [below](#x-ck12-Qy1CaW8tMDMtMTEtRG93bi1zeW5kcm9t) ) |
| Turner’s syndrome | one X chromosome but no other sex chromosome (XO) | female with short height and infertility (inability to reproduce) |
| Triple X syndrome | three X chromosomes (XXX) | female with mild developmental delays and menstrual irregularities |
| Klinefelter’s syndrome | one Y chromosome and two or more X chromosomes (XXY, XXXY) | male with problems in sexual development and reduced levels of the male hormone testosterone |

(left) Trisomy 21 (Down Syndrome) Karyotype. A **karyotype** is a picture of a cell's chromosomes. Note the extra chromosome 21. (right) Child with Down syndrome, exhibiting characteristic facial appearance.

#### Diagnosing Genetic Disorders

A genetic disorder that is caused by a mutation can be inherited. Therefore, people with a genetic disorder in their family may be concerned about having children with the disorder. Professionals known as **genetic counselors** can help them understand the risks of their children being affected. If they decide to have children, they may be advised to have **prenatal** (“before birth”) testing to see if the fetus has any genetic abnormalities. One method of prenatal testing is **amniocentesis**. In this procedure, a few fetal cells are extracted from the fluid surrounding the fetus, and the fetal chromosomes are examined.

#### Treating Genetic Disorders

The symptoms of genetic disorders can sometimes be treated, but cures for genetic disorders are still in the early stages of development. One potential cure that has already been used with some success is **gene therapy**. This involves inserting normal genes into cells with mutant genes. At the following link, you can watch the video "Sickle Cell Anemia: Hope from Gene Therapy," to learn how scientists are trying to cure sickle-cell anemia with gene therapy. <http://www.pubinfo.vcu.edu/secretsofthesequence/playlist_frame.asp>

If you could learn your risk of getting cancer or another genetic disease, would you? Though this is a personal decision, it is a possibility. A San Francisco company now makes it easy to order medical genetic tests through the Web. See "Genetic Testing through the Web" at <http://www.kqed.org/quest/television/genetic-testing-through-the-web>.

### Summary

* Many genetic disorders are caused by mutations in one or a few genes.
* Other genetic disorders are caused by abnormal numbers of chromosomes.

### Practice

Use this resource to answer the questions that follow.

* <http://www.hippocampus.org/Biology>Biology for AP\* Search: **Chromosomal Abnormalities**

1. What is chromosome nondisjunction?
2. What is Turner syndrome? What are its characteristics?
3. Describe the outcomes of chromosome nondisjunction in a male.
4. Define aneuploidy.
5. What is the difference between monosomy and trisomy?
6. What are the odds that a 45-year-old woman will have a baby with Down syndrome?
7. What is a karyotype? What can a karyotype demonstrate?
8. What is AML? What can cause AML?

# 4.22 Biotechnology

* Describe gene cloning and the polymerase chain reaction.

**So how does a scientist work with DNA?**

It always starts with the sequence. Once the sequence is known, so much more can be done. Specific regions can be isolated, cloned, amplified, and then used to help us.

### Biotechnology Methods

**Biotechnology** is the use of technology to change the genetic makeup of living things for human purposes. Generally, the purpose of biotechnology is to create organisms that are useful to humans or to cure genetic disorders. For example, biotechnology may be used to create crops that resist insect pests or yield more food, or to create new treatments for human diseases.

Biotechnology: The Invisible Revolution can be seen at <http://www.youtube.com/watch?v=OcG9q9cPqm4>.

What does biotechnology have to do with me? is discussed in the following video: <http://www.youtube.com/watch?v=rrT5BT_7HdI&feature=related>(10:01).

Biotechnology uses a variety of techniques to achieve its aims. Two commonly used techniques are gene cloning and the polymerase chain reaction.

#### Gene Cloning

**Gene cloning** is the process of isolating and making copies of a gene. This is useful for many purposes. For example, gene cloning might be used to isolate and make copies of a normal gene for gene therapy. Gene cloning involves four steps: isolation, ligation, transformation, and selection. You can watch an interactive animation about gene cloning at this link: <http://www.teachersdomain.org/asset/biot09_int_geneclone/>.

1. In isolation, an enzyme (called a restriction enzyme) is used to break DNA at a specific base sequence. This is done to isolate a gene.
2. During **ligation**, the enzyme **DNA ligase** combines the isolated gene with plasmid DNA from bacteria. (A **plasmid** is circular DNA apart from the chromosome and can replicate independently.) Ligation is illustrated in **Figure** [below](#x-ck12-QmlvLTA4LTE0LWxpZ2F0aW9u) . The DNA that results is called **recombinant DNA**.
3. In **transformation**, the recombinant DNA is inserted into a living cell, usually a bacterial cell. Changing an organism in this way is also called **genetic engineering**.
4. Selection involves growing transformed bacteria to make sure they have the recombinant DNA. This is a necessary step because transformation is not always successful. Only bacteria that contain the recombinant DNA are selected for further use.

Ligation. DNA ligase joins together an isolated gene and plasmid DNA. This produces recombinant DNA.

Recombinant DNA technology is discussed in the following videos and animations: <http://www.youtube.com/watch?v=x2jUMG2E-ic>(4.36), <http://www.youtube.com/watch?v=Jy15BWVxTC0>(0.50), <http://www.youtube.com/watch?v=sjwNtQYLKeU&feature=related>(7.20), <http://www.youtube.com/watch?v=Fi63VjfhsfI>(3:59).

#### Polymerase Chain Reaction

The **polymerase chain reaction (PCR)** makes many copies of a gene or other DNA segment. This might be done in order to make large quantities of a gene for genetic testing. PCR involves three steps: **denaturing**, **annealing**, and **extension**. The three steps are illustrated in **Figure** [below](#x-ck12-QmlvLTA4LTE1LVBvbHltZXJhc2UtY2hh) . They are repeated many times in a cycle to make large quantities of the gene. You can watch animations of PCR at these links:

* <http://www.dnalc.org/resources/3d/19-polymerase-chain-reaction.html>
* <http://www.teachersdomain.org/asset/biot09_int_pcr/>.

1. Denaturing involves heating DNA to break the bonds holding together the two DNA strands. This yields two single strands of DNA.
2. Annealing involves cooling the single strands of DNA and mixing them with short DNA segments called **primers**. Primers have base sequences that are complementary to segments of the single DNA strands. As a result, bonds form between the DNA strands and primers.
3. Extension occurs when an enzyme ( **Taq polymerase** or Taq DNA polymerase) adds nucleotides to the primers. This produces new DNA molecules, each incorporating one of the original DNA strands.

The Polymerase Chain Reaction. The polymerase chain reaction involves three steps. High temperatures are needed for the process to work. The enzyme Taq polymerase is used in step 3 because it can withstand high temperatures.

### Summary

* Biotechnology is the use of technology to change the genetic makeup of living things for human purposes.
* Gene cloning is the process of isolating and making copies of a DNA segment such as a gene.
* The polymerase chain reaction makes many copies of a gene or other DNA segment.

### Practice

Use this resource and the videos associated with this resource to answer the questions that follow.

* **Polymerase Chain Reaction** at <http://www.dnalc.org/resources/spotlight/index.html>.

1. Who developed PCR?
2. What does PCR allow?
3. Describe the 3 steps involved in PCR.
4. Approximately how many copies of a specific segment of DNA can be made by PCR?

# 4.23 Biotechnology Applications

* Explain how DNA technology is applied in medicine and agriculture.

**Why would anyone grow plants like this?**

Developing better crops is a significant aspect of biotechnology. Crops that are resistant to damage from insects or droughts must have a significant role in the world's future. And it all starts in the lab.

### Applications of Biotechnology

Methods of biotechnology can be used for many practical purposes. They are used widely in both medicine and agriculture. To see how biotechnology can be used to solve crimes, watch the video "Justice DNA—Freeing the Innocent" at the following link: <http://www.pubinfo.vcu.edu/secretsofthesequence/playlist_frame.asp>.

#### Applications in Medicine

In addition to gene therapy for genetic disorders, biotechnology can be used to transform bacteria so they are able to make human proteins. **Figure** [below](#x-ck12-QmlvLTA4LTE2LWdlbmV0aWMtRW5naW5l) shows how this is done to produce a **cytokine**, which is a small protein that helps fight infections. Proteins made by the bacteria are injected into people who cannot produce them because of mutations.

Genetically Engineering Bacteria to Produce a Human Protein. Bacteria can be genetically engineered to produce a human protein, such as a cytokine. A cytokine is a small protein that helps fight infections.

**Insulin** was the first human protein to be produced in this way. Insulin helps cells take up glucose from the blood. People with type 1 diabetes have a mutation in the gene that normally codes for insulin. Without insulin, their blood glucose rises to harmfully high levels. At present, the only treatment for type 1 diabetes is the injection of insulin from outside sources. Until recently, there was no known way to make insulin outside the human body. The problem was solved by gene cloning. The human insulin gene was cloned and used to transform bacterial cells, which could then produce large quantities of human insulin.

#### Pharmacogenomics

We know that, thanks to our DNA, each of us is a little bit different. Some of those differences are obvious, like eye and hair color. Others are not so obvious, like how our bodies react to medication. Researchers are beginning to look at how to tailor medical treatments to our genetic profiles, in a relatively new field called **pharmacogenomics**. Some of the biggest breakthroughs have been in cancer treatment. For additional information on this “personalized medicine,” listen to <http://www.kqed.org/quest/radio/personalized-medicine>and see <http://www.kqed.org/quest/blog/2009/09/11/reporters-notes-personalized-medicine/>.

#### Synthetic Biology

Imagine living cells acting as memory devices, biofuels brewing from yeast, or a light receptor taken from algae that makes photographs on a plate of bacteria. The new field of **synthetic biology** is making biology easier to engineer so that new functions can be derived from living systems. Find out the tools that synthetic biologists are using and the exciting things they are building at <http://www.kqed.org/quest/television/decoding-synthetic-biology>and <http://www.kqed.org/quest/television/web-extra-synthetic-biology-extended-interview>.

Go to <http://goo.gl/zVthrf> for more content

#### Applications in Agriculture

Biotechnology has been used to create transgenic crops. **Transgenic crops** are genetically modified with new genes that code for traits useful to humans. The diagram in **Figure** [below](#x-ck12-QmlvLTA4LTE3LVRyYW5zZ2VuaWMtY3Jv) shows how a transgenic crop is created. You can learn more about how scientists create transgenic crops with the interactive animation "Engineer a Crop: Transgenic Manipulation" at this link: <http://www.pbs.org/wgbh/harvest/engineer/transgen.html>.

Creating a Transgenic Crop. A transgenic crop is genetically modified to be more useful to humans.

Transgenic crops have been created with a variety of different traits, such as yielding more food, tasting better, surviving drought, and resisting insect pests. Scientists have even created a transgenic purple tomato that contains a cancer-fighting compound (see **Figure** [below](#x-ck12-QmlvLTA4LTE4LVB1cnBsZS10cmFuc2dl) ). To learn how scientists have used biotechnology to create plants that can grow in salty soil, watch the video "Salt of the Earth - Engineering Salt-tolerant Plants" at this link: <http://www.sosq.vcu.edu/videos.aspx>.

Transgenic Purple Tomato. A purple tomato is genetically modified to contain a cancer-fighting compound. A gene for the compound was transferred into normal red tomatoes.

Biotechnology in agriculture is discussed at <http://www.youtube.com/watch?v=IY3mfgbe-0c>(6:40).

### Summary

* Biotechnology can be used to transform bacteria so they are able to make human proteins, such as insulin.
* It can also be used to create transgenic crops, such as crops that yield more food or resist insect pests.

### Practice I

Use these resources to answer the questions that follow.

* **Craig Venter** at <http://www.tedmed.com/videos-info?name=Craig_Venter_at_TEDMED_2010&q=updated&year=all>

Go to <http://goo.gl/eqkaSC> for more content

1. How can biotechnology help with agricultural issues?
2. What is test-tube cloning?
3. Describe Golden rice.
4. Describe how advances in biotechnology have helped medical applications.
5. What is a DNA fingerprint? Does every person have an unique DNA fingerprint?
6. How is a DNA fingerprint used?

# 4.24 Ethical, Legal, and Social Issues of Biotechnology

* Identify some of the ethical, legal, and social issues raised by biotechnology.

**Right or wrong? Good or bad? Legal or illegal?**

The completion of The Human Genome Project is one of the most important scientific events of the past 50 years. However, is knowing all of our DNA a good thing? The advancement of biotechnology has raised many interesting ethical, legal and social questions.

### Ethical, Legal, and Social Issues

Imagine someone analyzes part of your DNA. Who controls that information? What if your health insurance company found out you were predisposed to develop a devastating genetic disease. Might they decide to cancel your insurance? Privacy issues concerning genetic information is an important issue in this day and age.

**ELSI** stands for Ethical, Legal and Social Issues. It's a term associated with the Human Genome project. This project didn't only have the goal to identify all the genes in the human genome, but also to address the ELSI that might arise from the project. Rapid advances in DNA-based research, human genetics, and their applications have resulted in new and complex ethical and legal issues for society.

#### Concerns from Biotechnology

The use of biotechnology has raised a number of ethical, legal, and social issues. Here are just a few:

* Who owns genetically modified organisms such as bacteria? Can such organisms be patented like inventions?
* Are genetically modified foods safe to eat? Might they have unknown harmful effects on the people who consume them?
* Are genetically engineered crops safe for the environment? Might they harm other organisms or even entire ecosystems?
* Who controls a person’s genetic information? What safeguards ensure that the information is kept private?
* How far should we go to ensure that children are free of mutations? Should a pregnancy be ended if the fetus has a mutation for a serious genetic disorder?

Addressing such issues is beyond the scope of this concept. The following example shows how complex the issues may be:

A strain of corn has been created with a gene that encodes a natural pesticide. On the positive side, the **transgenic** corn is not eaten by insects, so there is more corn for people to eat. The corn also doesn’t need to be sprayed with chemical pesticides, which can harm people and other living things. On the negative side, the transgenic corn has been shown to cross-pollinate nearby milkweed plants. Offspring of the cross-pollinated milkweed plants are now known to be toxic to monarch butterfly caterpillars that depend on them for food. Scientists are concerned that this may threaten the monarch species as well as other species that normally eat monarchs.

As this example shows, the pros of biotechnology may be obvious, but the cons may not be known until it is too late. Unforeseen harm may be done to people, other species, and entire ecosystems. No doubt the ethical, legal, and social issues raised by biotechnology will be debated for decades to come. For a recent debate about the ethics of applying biotechnology to humans, watch the video at the link below. In the video, a Harvard University professor of government and a Princeton University professor of bioethics debate the science of “perfecting humans.” <http://www.youtube.com/watch?v=-BPna-fSNOE>

### Summary

* Biotechnology has raised a number of ethical, legal, and social issues. For example, are genetically modified foods safe to eat, and who controls a person’s genetic information?

### Practice

Use this resource to answer the questions that follow.

* <http://www.hippocampus.org/Biology>Biology for AP\* Search: **Practical & Ethical Concerns**

1. What are two concerns associated with biotechnology?
2. Why could genetically engineered plants replace naturally grown plants?
3. What is cloning? What was the first cloned large mammal?
4. What are two ethical considerations associated with the human genome sequence?

## Summary

Beginning with Mendel's pea plants, genetics has become one of the most important fields of biology. Genetics discusses genetics, from Mendel's pea plants to current ethical issues associated with this field. The completion of The Human Genome Project is one of the landmark scientific events of the last 50 years. Human genetics affects many, if not every, field of medicine. Technologies associated with genetics are involved in developing products to make our lives better, but have raised a number of ethical, legal and social issues.

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