



# Help Me Understand Genetics

## Cells and DNA

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# Cells and DNA

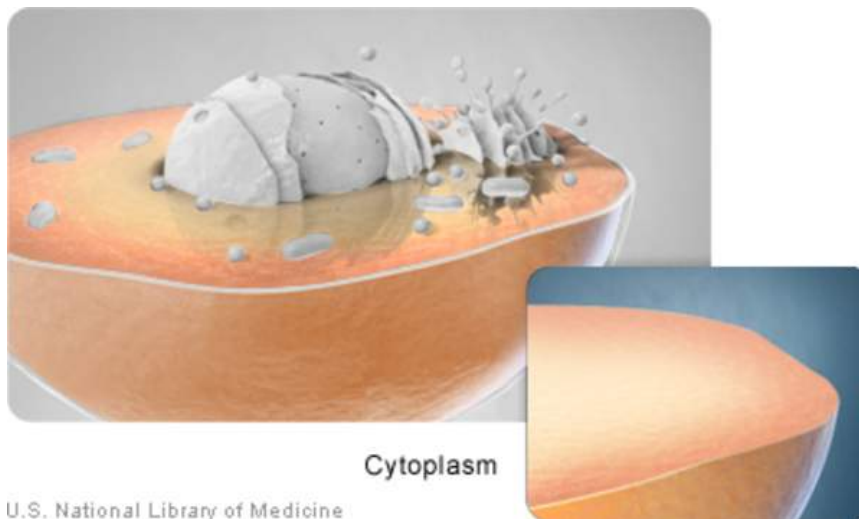
## 1 What is a cell?

Cells are the basic building blocks of all living things. The human body is composed of trillions of cells. They provide structure for the body, take in nutrients from food, convert those nutrients into energy, and carry out specialized functions. Cells also contain the body's hereditary material and can make copies of themselves.

Cells have many parts, each with a different function. Some of these parts, called organelles, are specialized structures that perform certain tasks within the cell. Human cells contain the following major parts, listed in alphabetical order:

### **Cytoplasm**

Within cells, the cytoplasm (Figure 1) is made up of a jelly-like fluid (called the cytosol) and other structures that surround the nucleus.



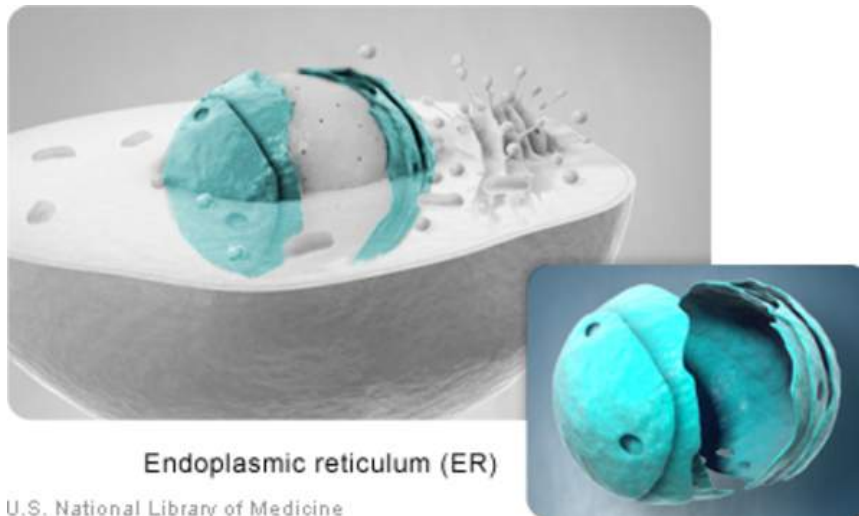
**FIGURE 1:** The cytoplasm is fluid within cells that surrounds the nucleus.

### **Cytoskeleton**

The cytoskeleton is a network of long fibers that make up the cell's structural framework. The cytoskeleton has several critical functions, including determining cell shape, participating in cell division, and allowing cells to move. It also provides a track-like system that directs the movement of organelles and other substances within cells.

### **Endoplasmic reticulum (ER)**

This organelle helps process molecules created by the cell. The endoplasmic reticulum (Figure 2) also transports these molecules to their specific destinations either inside or outside the cell.



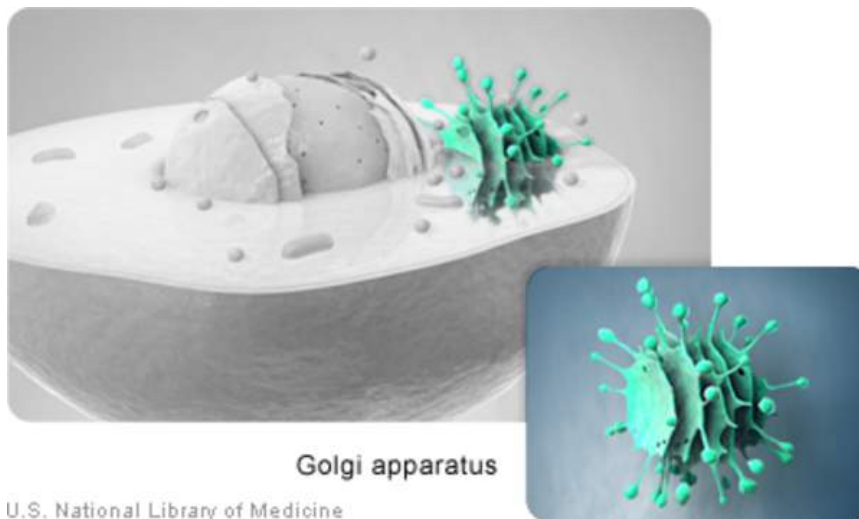
Endoplasmic reticulum (ER)

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**FIGURE 2:** The endoplasmic reticulum is a structure near the cell nucleus.

### **Golgi apparatus**

The Golgi apparatus (Figure 3) packages molecules processed by the endoplasmic reticulum to be transported out of the cell.



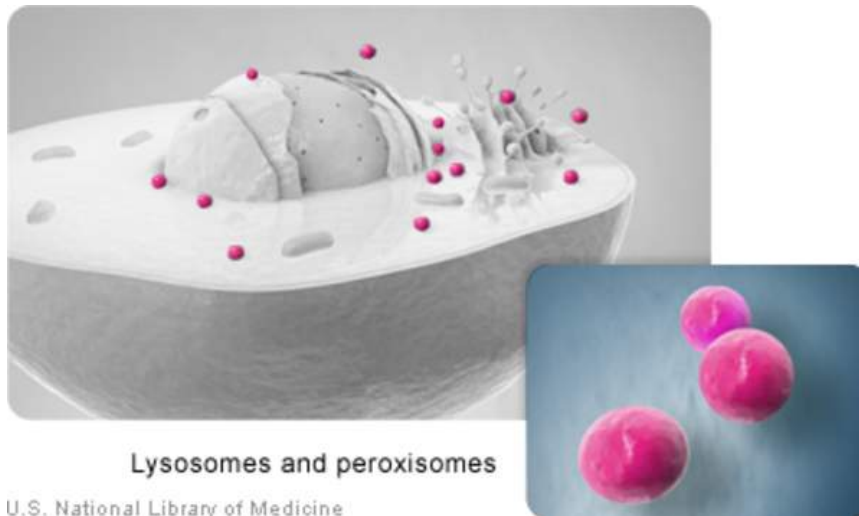
Golgi apparatus

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**FIGURE 3:** The Golgi apparatus is a structure in the cell cytoplasm.

### **Lysosomes and peroxisomes**

These organelles (Figure 4) are the recycling center of the cell. They digest foreign bacteria that invade the cell, rid the cell of toxic substances, and recycle worn-out cell components.



Lysosomes and peroxisomes

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**FIGURE 4:** Lysosomes are cell structures in the cytoplasm.

## Mitochondria

Mitochondria (Figure 5) are complex organelles that convert energy from food into a form that the cell can use. They have their own genetic material, separate from the DNA in the nucleus, and can make copies of themselves.



Mitochondria

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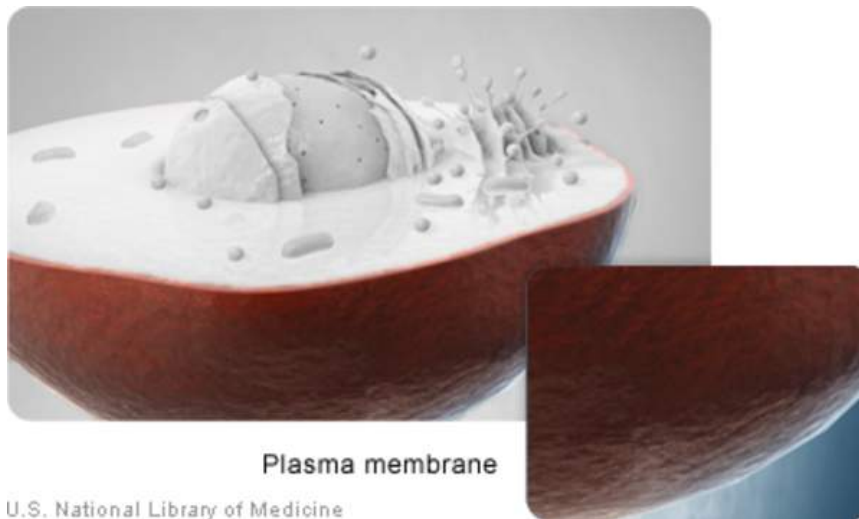
**FIGURE 5:** Mitochondria are structures within the cell cytoplasm.

## Nucleus

The nucleus serves as the cell's command center, sending directions to the cell to grow, mature, divide, or die. It also houses DNA (deoxyribonucleic acid), the cell's hereditary material. The nucleus is surrounded by a membrane called the nuclear envelope, which protects the DNA and separates the nucleus from the rest of the cell.

## Plasma membrane

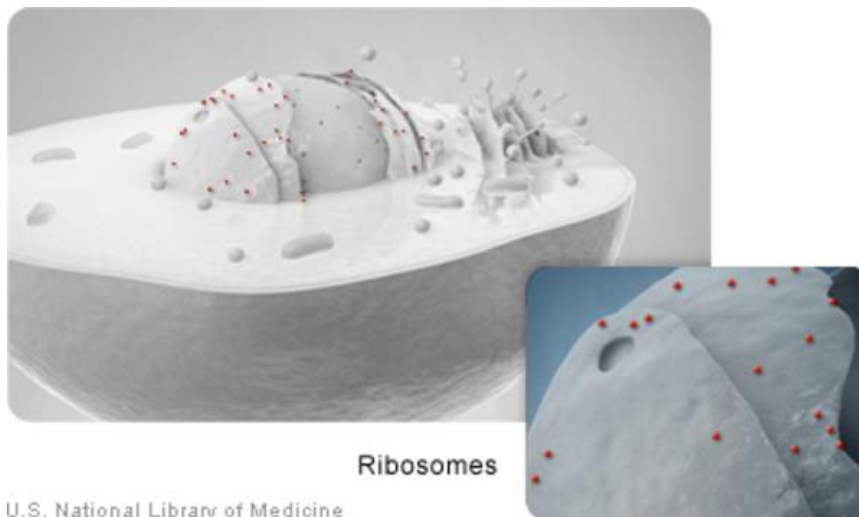
The plasma membrane (Figure 6) is the outer lining of the cell. It separates the cell from its environment and allows materials to enter and leave the cell.



**FIGURE 6:** The plasma membrane surrounds a cell.

## Ribosomes

Ribosomes (Figure 7) are organelles that process the cell's genetic instructions to create proteins. These organelles can float freely in the cytoplasm or be connected to the endoplasmic reticulum (see above).



**FIGURE 7:** Ribosomes are cell structures near the nucleus.

**For more information about cells:**

The National Institute of General Medical Sciences has a science education booklet about cells called Inside the Cell (<https://www.nigms.nih.gov/education/Booklets/Inside-the-Cell/Pages/Home.aspx>).

The National Human Genome Research Institute's Talking Glossary (<https://www.genome.gov/genetics-glossary/Cell>) provides a definition of a cell as well as an illustration and video.

The Genetic Science Learning Center at the University of Utah offers an interactive introduction to cells (<https://learn.genetics.utah.edu/content/cells/insideacell/>) and their many functions.

Arizona State University's "Ask a Biologist" provides a description and illustration of each of the cell's organelles (<https://askabiologist.asu.edu/cell-parts>).

Queen Mary University of London allows you to explore a 3-D cell and its parts (<https://www.centreofthecell.org/learn-play/games/explore-a-cell/>).

## 2 What is DNA?

DNA, or deoxyribonucleic acid, is the hereditary material in humans and almost all other organisms. Nearly every cell in a person's body has the same DNA. Most DNA is located in the cell nucleus (where it is called nuclear DNA), but a small amount of DNA can also be found in the mitochondria (where it is called mitochondrial DNA or mtDNA). Mitochondria (Figure 5) are structures within cells that convert the energy from food into a form that cells can use.

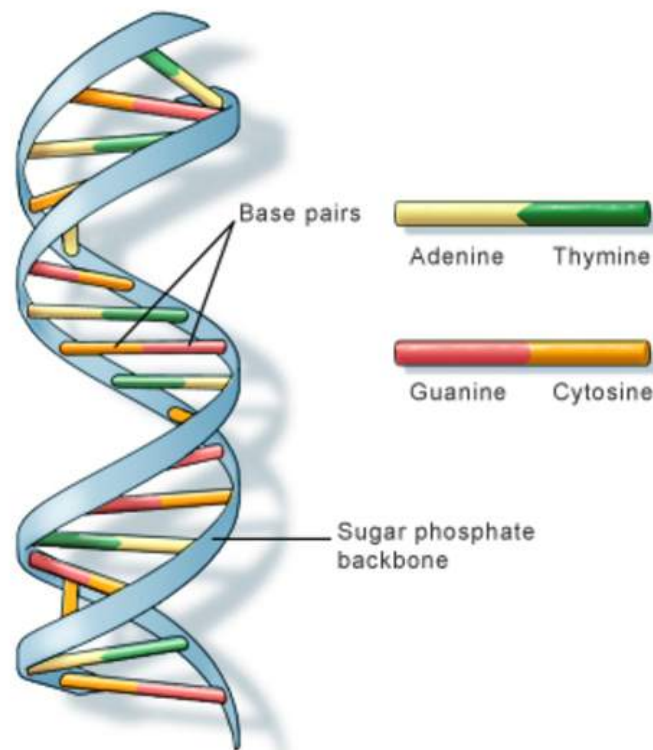
The information in DNA is stored as a code made up of four chemical bases: adenine (A), guanine (G), cytosine (C), and thymine (T). Human DNA consists of about 3 billion bases, and more than 99 percent of those bases are the same in all people. The order, or sequence, of these bases determines the information available for building and maintaining an organism, similar to the way in which letters of the alphabet appear in a certain order to form words and sentences.

DNA bases pair up with each other, A with T and C with G, to form units called base pairs. Each base is also attached to a sugar molecule and a phosphate molecule. Together, a base, sugar, and phosphate are called a nucleotide. Nucleotides are arranged in two long strands that form a spiral called a double helix. The structure of the double helix is somewhat like a ladder, with the base pairs forming the ladder's rungs and the sugar and phosphate molecules forming the vertical sidepieces of the ladder.

An important property of DNA is that it can replicate, or make copies of itself. Each strand of DNA in the double helix can serve as a pattern for duplicating the sequence of bases. This is critical when cells divide because each new cell needs to have an exact copy of the DNA present in the old cell.

DNA is a double helix formed by base pairs attached to a sugar-phosphate backbone. (Figure 8)





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**FIGURE 8:** DNA is made up of base pairs and a sugar phosphate backbone.

### For more information about DNA:

The National Human Genome Research Institute (NHGRI), a part of the National Institutes of Health (NIH), provides a fact sheet Deoxyribonucleic Acid (DNA) (<https://www.genome.gov/about-genomics/fact-sheets/Deoxyribonucleic-Acid-Fact-Sheet>) that gives an introduction to this molecule.

StatedClearly offers a video introduction to DNA and how it works (<https://www.statedclearly.com/videos/what-is-dna/>).

The New Genetics, a publication of the National Institute of General Medical Sciences, part of the NIH, discusses the structure of DNA and how it was discovered (<https://www.nigms.nih.gov/education/Booklets/the-new-genetics/Pages/Home.aspx>).

A basic explanation and illustration of DNA (<https://askabiologist.asu.edu/dna-shape-and-structure>) can be found on Arizona State University's "Ask a Biologist" website.

The Virtual Genetics Education Centre, created by the University of Leicester, offers

additional information on DNA, genes, and chromosomes (<https://www2.le.ac.uk/projects/vgec/highereducation/topics/dna-genes-chromosomes>).

An overview of mitochondrial DNA (<https://neuromuscular.wustl.edu/mitosyn.html#general>) is available from the Neuromuscular Disease Center at Washington University in St. Louis.

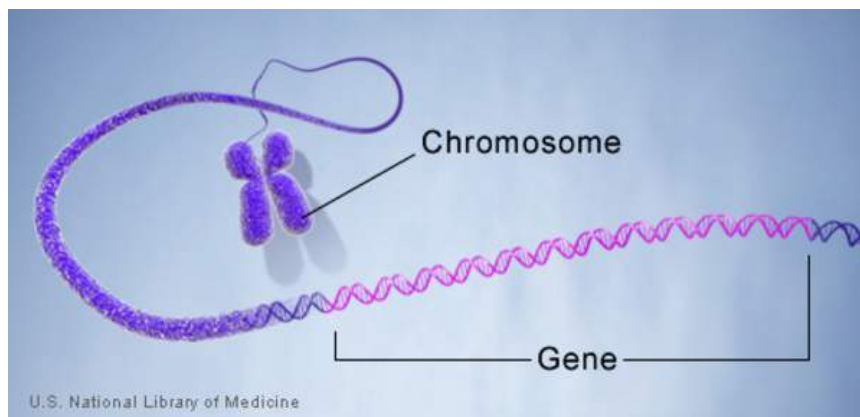
### 3 What is a gene?

A gene is the basic physical and functional unit of heredity. Genes are made up of DNA. Some genes act as instructions to make molecules called proteins. However, many genes do not code for proteins. In humans, genes vary in size from a few hundred DNA bases to more than 2 million bases. An international research effort called the Human Genome Project, which worked to determine the sequence of the human genome and identify the genes that it contains, estimated that humans have between 20,000 and 25,000 genes.

Every person has two copies of each gene, one inherited from each parent. Most genes are the same in all people, but a small number of genes (less than 1 percent of the total) are slightly different between people. Alleles are forms of the same gene with small differences in their sequence of DNA bases. These small differences contribute to each person's unique physical features.

Scientists keep track of genes by giving them unique names. Because gene names can be long, genes are also assigned symbols, which are short combinations of letters (and sometimes numbers) that represent an abbreviated version of the gene name. For example, a gene on chromosome 7 that has been associated with cystic fibrosis is called the cystic fibrosis transmembrane conductance regulator; its symbol is *CFTR*.

Genes are made up of DNA. Each chromosome contains many genes. (Figure 9)



**FIGURE 9:** A gene is labeled along the length of a chromosome.

#### **For more information about genes:**

MedlinePlus Genetics provides consumer-friendly gene summaries (<https://medlineplus.gov/genetics/gene/>) that include an explanation of each gene's normal function and how variants in the gene cause particular genetic conditions.

More information about how genetic conditions and genes are named (<https://medlineplus.gov/genetics/understanding/mutationsanddisorders/naming/>) is also available from MedlinePlus Genetics.

The Centre for Genetics Education offers a fact sheet that introduces genes and chromosomes (<https://www.genetics.edu.au/SitePages/DNA-RNA-genes-and-chromosomes.aspx>).

The Tech Museum of Innovation at Stanford University describes genes and how they were discovered (<https://genetics.thetech.org/about-genetics/what-gene>).

The Virtual Genetics Education Centre, created by the University of Leicester, offers additional information on DNA, genes, and chromosomes (<https://www2.le.ac.uk/projects/vgec/highereducation/topics/dna-genes-chromosomes>).

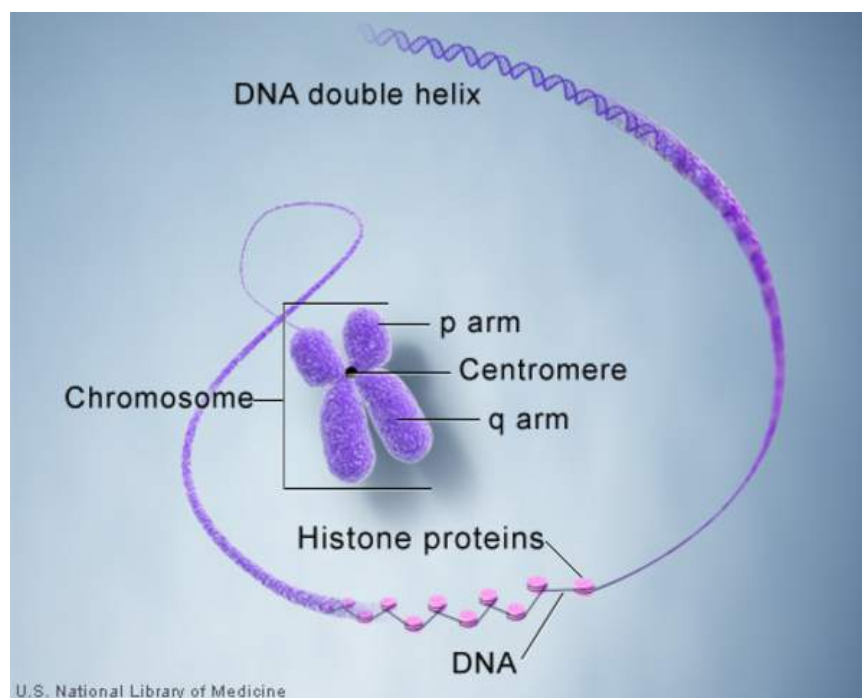
## 4 What is a chromosome?

In the nucleus of each cell, the DNA molecule is packaged into thread-like structures called chromosomes. Each chromosome is made up of DNA tightly coiled many times around proteins called histones that support its structure.

Chromosomes are not visible in the cell's nucleus—not even under a microscope—when the cell is not dividing. However, the DNA that makes up chromosomes becomes more tightly packed during cell division and is then visible under a microscope. Most of what researchers know about chromosomes was learned by observing chromosomes during cell division.

Each chromosome has a constriction point called the centromere, which divides the chromosome into two sections, or “arms.” The short arm of the chromosome is labeled the “p arm.” The long arm of the chromosome is labeled the “q arm.” The location of the centromere on each chromosome gives the chromosome its characteristic shape, and can be used to help describe the location of specific genes.

DNA and histone proteins are packaged into structures called chromosomes. (Figure 10)



**FIGURE 10:** Chromosomes have a p arm, a q arm, and a centromere. They are made up of DNA wrapped around histone proteins.

### **For more information about chromosomes:**

MedlinePlus Genetics provides information about each human chromosome (<https://medlineplus.gov/genetics/chromosome/>) written in lay language.

A basic introduction to chromosomes (<https://www.genome.gov/about-genomics/fact-sheets/Chromosomes-Fact-Sheet>) is available from the National Human Genome Research Institute.

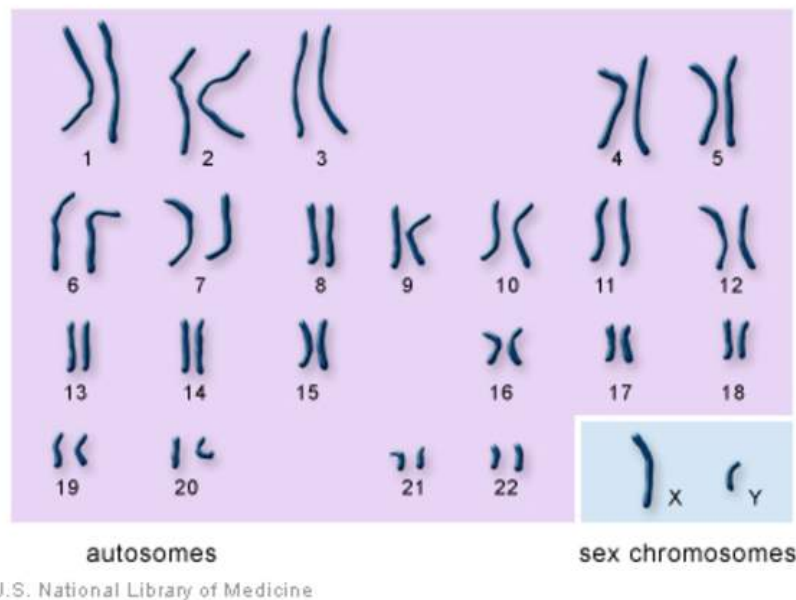
The Centre for Genetics Education offers a fact sheet that introduces genes and chromosomes (<https://www.genetics.edu.au/SitePages/DNA-RNA-genes-and-chromosomes.aspx>).

The University of Utah's Genetic Science Learning Center offers a description of chromosomes (<https://learn.genetics.utah.edu/content/basics/readchromosomes/>), including how scientists tell them apart.

## 5 How many chromosomes do people have?

In humans, each cell normally contains 23 pairs of chromosomes, for a total of 46. Twenty-two of these pairs, called autosomes, look the same in both males and females. The 23rd pair, the sex chromosomes, differ between males and females. Females have two copies of the X chromosome, while males have one X and one Y chromosome.

The 22 autosomes are numbered by size. The other two chromosomes, X and Y, are the sex chromosomes. This picture of the human chromosomes lined up in pairs is called a karyotype. (Figure 11)



**FIGURE 11:** A normal male karyotype with 22 autosomes, one X chromosome, and one Y chromosome.

### For more information about the 23 pairs of human chromosomes:

MedlinePlus Genetics provides information about each human chromosome (<https://medlineplus.gov/genetics/chromosome/>) written in lay language.

MedlinePlus offers additional details about karyotype genetic tests (<https://medlineplus.gov/lab-tests/karyotype-genetic-test/>).

The University of Utah's Genetic Science Learning Center discusses how karyotypes can be used in diagnosing genetic disorders (<https://learn.genetics.utah.edu/content/basics/diagnose/>).

Arizona State University's "Ask a Biologist" discusses the inheritance of human

chromosomes. (<https://askbiologist.asu.edu/chromosomes-and-genes>)



## 6 What is noncoding DNA?

Only about 1 percent of DNA is made up of protein-coding genes; the other 99 percent is noncoding. Noncoding DNA does not provide instructions for making proteins. Scientists once thought noncoding DNA was “junk,” with no known purpose. However, it is becoming clear that at least some of it is integral to the function of cells, particularly the control of gene activity. For example, noncoding DNA contains sequences that act as regulatory elements, determining when and where genes are turned on and off. Such elements provide sites for specialized proteins (called transcription factors) to attach (bind) and either activate or repress the process by which the information from genes is turned into proteins (transcription). Noncoding DNA contains many types of regulatory elements:

- Promoters provide binding sites for the protein machinery that carries out transcription. Promoters are typically found just ahead of the gene on the DNA strand.
- Enhancers provide binding sites for proteins that help activate transcription. Enhancers can be found on the DNA strand before or after the gene they control, sometimes far away.
- Silencers provide binding sites for proteins that repress transcription. Like enhancers, silencers can be found before or after the gene they control and can be some distance away on the DNA strand.
- Insulators provide binding sites for proteins that control transcription in a number of ways. Some prevent enhancers from aiding in transcription (enhancer-blocker insulators). Others prevent structural changes in the DNA that repress gene activity (barrier insulators). Some insulators can function as both an enhancer blocker and a barrier.

Other regions of noncoding DNA provide instructions for the formation of certain kinds of RNA molecules. RNA is a chemical cousin of DNA. Examples of specialized RNA molecules produced from noncoding DNA include transfer RNAs (tRNAs) and ribosomal RNAs (rRNAs), which help assemble protein building blocks (amino acids) into a chain that forms a protein; microRNAs (miRNAs), which are short lengths of RNA that block the process of protein production; and long noncoding RNAs (lncRNAs), which are longer lengths of RNA that have diverse roles in regulating gene activity.

Some structural elements of chromosomes are also part of noncoding DNA. For example, repeated noncoding DNA sequences at the ends of chromosomes form telomeres. Telomeres protect the ends of chromosomes from being degraded during the copying of genetic material. Repetitive noncoding DNA sequences also form satellite DNA, which is a part of other structural elements. Satellite DNA is the basis of the centromere, which is the constriction point of the X-shaped chromosome pair. Satellite DNA also forms heterochromatin, which is densely packed DNA that is important for controlling gene activity and maintaining the structure of chromosomes.

Some noncoding DNA regions, called introns, are located within protein-coding genes but are removed before a protein is made. Regulatory elements, such as enhancers,

can be located in introns. Other noncoding regions are found between genes and are known as intergenic regions.

The identity of regulatory elements and other functional regions in noncoding DNA is not completely understood. Researchers are working to understand the location and role of these genetic components.

### **Scientific journal articles for further reading**

Maston GA, Evans SK, Green MR. Transcriptional regulatory elements in the human genome. *Annu Rev Genomics Hum Genet.* 2006;7:29-59. Review. PubMed: 16719718.

ENCODE Project Consortium. An integrated encyclopedia of DNA elements in the human genome. *Nature.* 2012 Sep 6;489(7414):57-74. doi: 10.1038/nature11247. PubMed: 22955616; Free full text available from PubMed Central: PMC3439153.

Plank JL, Dean A. Enhancer function: mechanistic and genome-wide insights come together. *Mol Cell.* 2014 Jul 3;55(1):5-14. doi: 10.1016/j.molcel.2014.06.015. Review. PubMed: 24996062.

### **For more information about noncoding DNA:**

Cold Spring Harbor Laboratory DNA Learning Center: The Human Genome: Genes and Non-coding DNA, 3D Animation with Basic Narration (<https://dnalc.cshl.edu/view/15486-The-human-genome-genes-and-non-coding-DNA-3D-animation-with-basic-narration.html>)

University of Leicester Virtual Genetics Education Centre: Gene Expression and Regulation (<https://www2.le.ac.uk/projects/vgec/highereducation/topics/geneexpression-regulation>)

National Academies Press: Noncoding DNA—Subtlety, Punctuation, or Just Plain Junk? (<https://www.nap.edu/read/1859/chapter/6#99>)

Khan Academy: Transcription Factors (<https://www.khanacademy.org/science/biology/gene-regulation/gene-regulation-in-eukaryotes/a/eukaryotic-transcription-factors>)

The Cell: A Molecular Approach (second edition, 2000): Regulation of Transcription in Eukaryotes (<https://www.ncbi.nlm.nih.gov/books/NBK9904/>)

Genetic Science Learning Center, University of Utah: RNA's Role in the Central Dogma (<https://learn.genetics.utah.edu/content/basics/centraldogma/>), Telomeres (<https://learn.genetics.utah.edu/content/basics/telomeres/>), and Centromeres (<https://learn.genetics.utah.edu/content/basics/readchromosomes/>)