Genetics Home Reference

Your Guide to Understanding Genetic Conditions

Handbook

Help Me Understand Genetics

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Department of Health & Human Services

Published November 12, 2013

Handbook

Table of Contents

Chapter 1

Cells and DNA

Table of Contents

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 (illustration on page 5)

Within cells, the cytoplasm is made up of a jelly-like fluid (called the cytosol) and other structures that surround 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) (illustration on page 6)

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

Golgi apparatus (illustration on page 6)

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

Lysosomes and peroxisomes (illustration on page 6)

These organelles 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.

Mitochondria (illustration on page 7)

Mitochondria 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.

Nucleus (illustration on page 7)

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 (illustration on page 7)

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

Ribosomes (illustration on page 8)

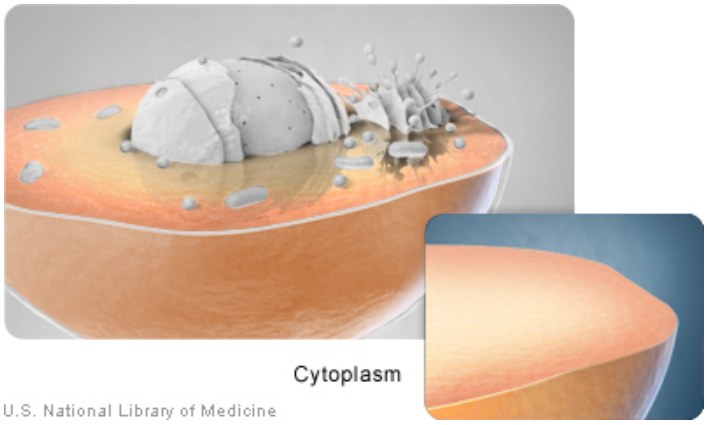
Ribosomes are organelles that process the cell's genetic instructions to create proteins. These organelles can float freely in the cytoplasm or be connected to the endoplasmic reticulum (see above).

For more information about cells:

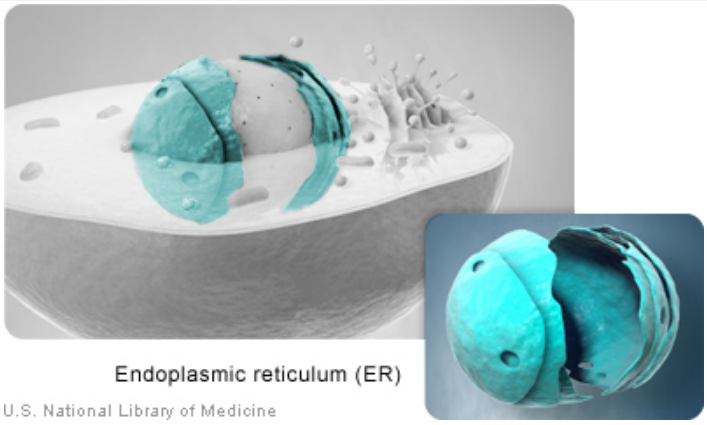
The Genetic Science Learning Center at the University of Utah offers an interactive introduction to cells (http://learn.genetics.utah.edu/content/begin/cells/) and their many functions.

Additional information about the cytoskeleton, including an illustration, is available from the Cytoplasm Tutorial (http://www.biology.arizona.edu/Cell_bio/tutorials/cytoskeleton/page1.html). This resource is part of The Biology Project at the University of Arizona.

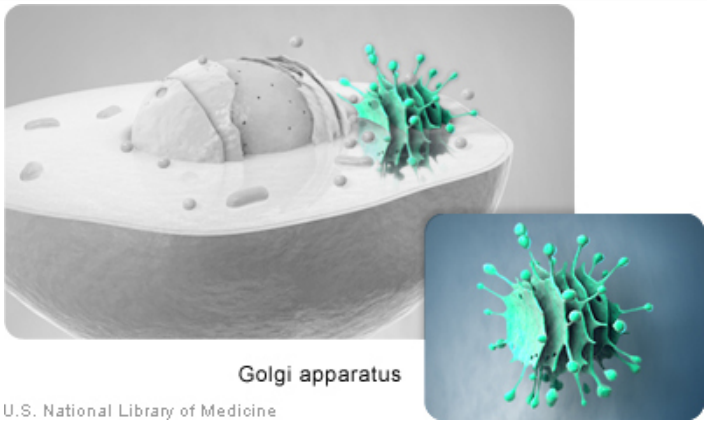
Illustrations



The cytoplasm surrounds the cell's nucleus and organelles.



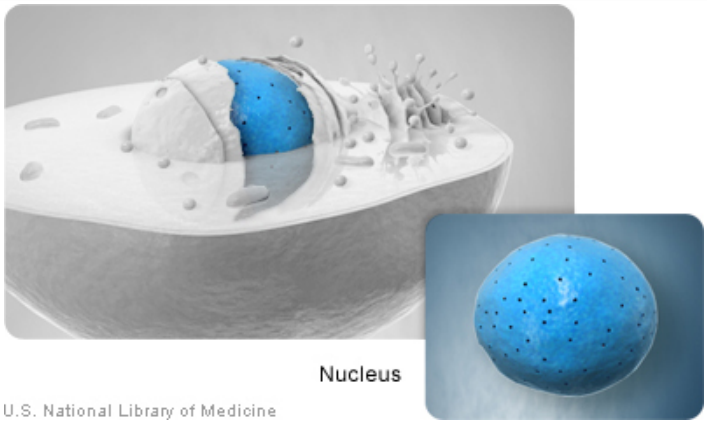
The endoplasmic reticulum is involved in molecule processing and transport.



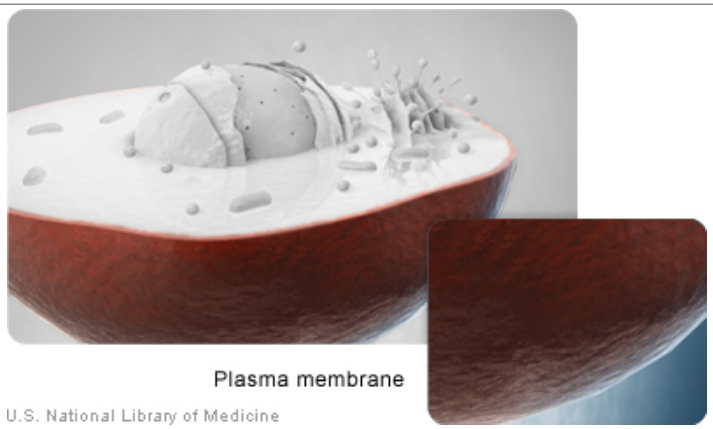
The Golgi apparatus is involved in packaging molecules for export from the cell.

Lysosomes and peroxisomes destroy toxic substances and recycle worn-out cell parts.

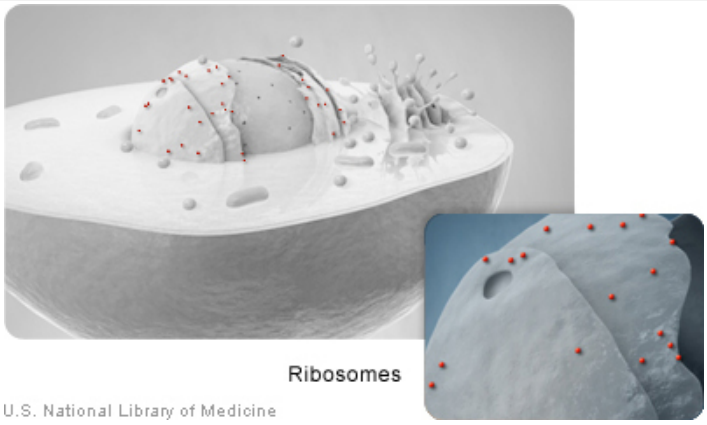
Mitochondria provide the cell's energy.



The nucleus contains most of the cell's genetic material.



The plasma membrane is the outer covering around the cell.



Ribosomes use the cell’s genetic instructions to make proteins.

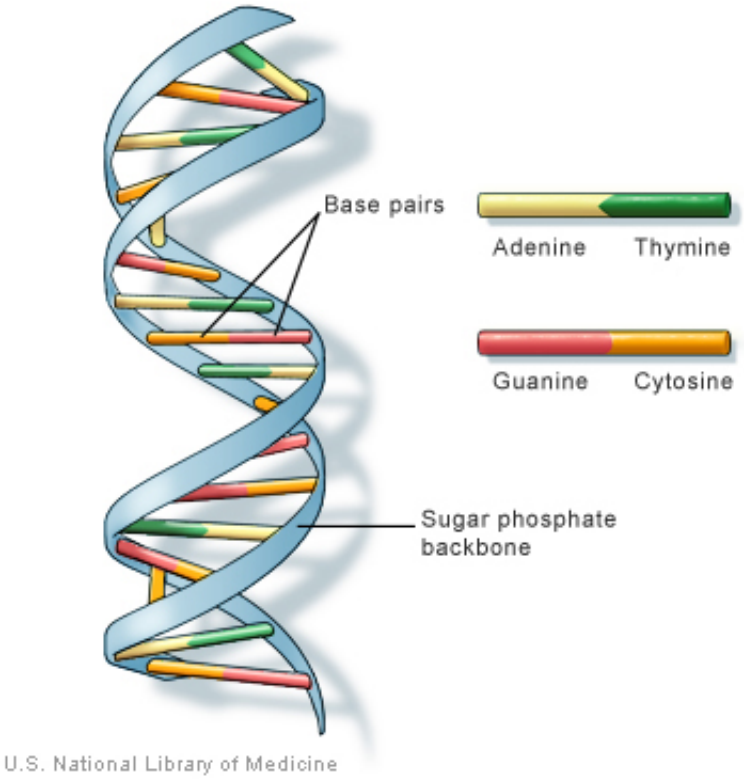
What is DNA?

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).

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.

For more information about DNA:

The National Human Genome Research Institute fact sheet Deoxyribonucleic Acid (DNA) (http://www.genome.gov/25520880) provides an introduction to this molecule.

Information about the genetic code (http://geneed.nlm.nih.gov/topic_subtopic.php?tid=15&sid=19) and the structure of the DNA double helix (http://geneed.nlm.nih.gov/topic_subtopic.php?tid=15&sid=16) is available from GeneEd.

The New Genetics, a publication of the National Institute of General Medical Sciences, discusses the structure of DNA and how it was discovered (http://publications.nigms.nih.gov/thenewgenetics/chapter1.html#c1).

What is mitochondrial DNA?

Although most DNA is packaged in chromosomes within the nucleus, mitochondria also have a small amount of their own DNA. This genetic material is known as mitochondrial DNA or mtDNA.

Mitochondria (illustration on page 7) are structures within cells that convert the energy from food into a form that cells can use. Each cell contains hundreds to thousands of mitochondria, which are located in the fluid that surrounds the nucleus (the cytoplasm).

Mitochondria produce energy through a process called oxidative phosphorylation. This process uses oxygen and simple sugars to create adenosine triphosphate (ATP), the cell's main energy source. A set of enzyme complexes, designated as complexes I-V, carry out oxidative phosphorylation within mitochondria.

In addition to energy production, mitochondria play a role in several other cellular activities. For example, mitochondria help regulate the self-destruction of cells (apoptosis). They are also necessary for the production of substances such as cholesterol and heme (a component of hemoglobin, the molecule that carries oxygen in the blood).

Mitochondrial DNA contains 37 genes, all of which are essential for normal mitochondrial function. Thirteen of these genes provide instructions for making enzymes involved in oxidative phosphorylation. The remaining genes provide instructions for making molecules called transfer RNAs (tRNAs) and ribosomal RNAs (rRNAs), which are chemical cousins of DNA. These types of RNA help assemble protein building blocks (amino acids) into functioning proteins.

For more information about mitochondria and mitochondrial DNA:

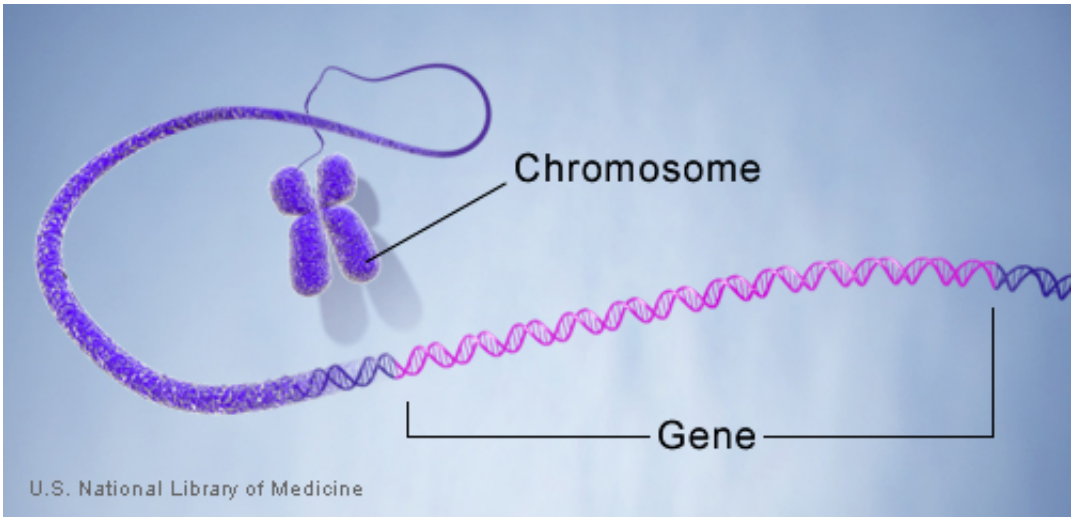
Molecular Expressions, a web site from the Florida State University Research Foundation, offers an illustrated introduction to mitochondria and mitochondrial DNA (http://micro.magnet.fsu.edu/cells/mitochondria/mitochondria.html).

An overview of mitochondrial DNA (http://neuromuscular.wustl.edu/mitosyn.html#general) is available from the Neuromuscular Disease Center at Washington University.

What is a gene?

A gene is the basic physical and functional unit of heredity. Genes, which are made up of DNA, act as instructions to make molecules called proteins. In humans, genes vary in size from a few hundred DNA bases to more than 2 million bases. The Human Genome Project has 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.



Genes are made up of DNA. Each chromosome contains many genes.

For more information about genes:

Genetics Home Reference provides consumer-friendly gene summaries (http://ghr.nlm.nih.gov/BrowseGenes) that include an explanation of each gene's normal function and how mutations in the gene cause particular genetic conditions.

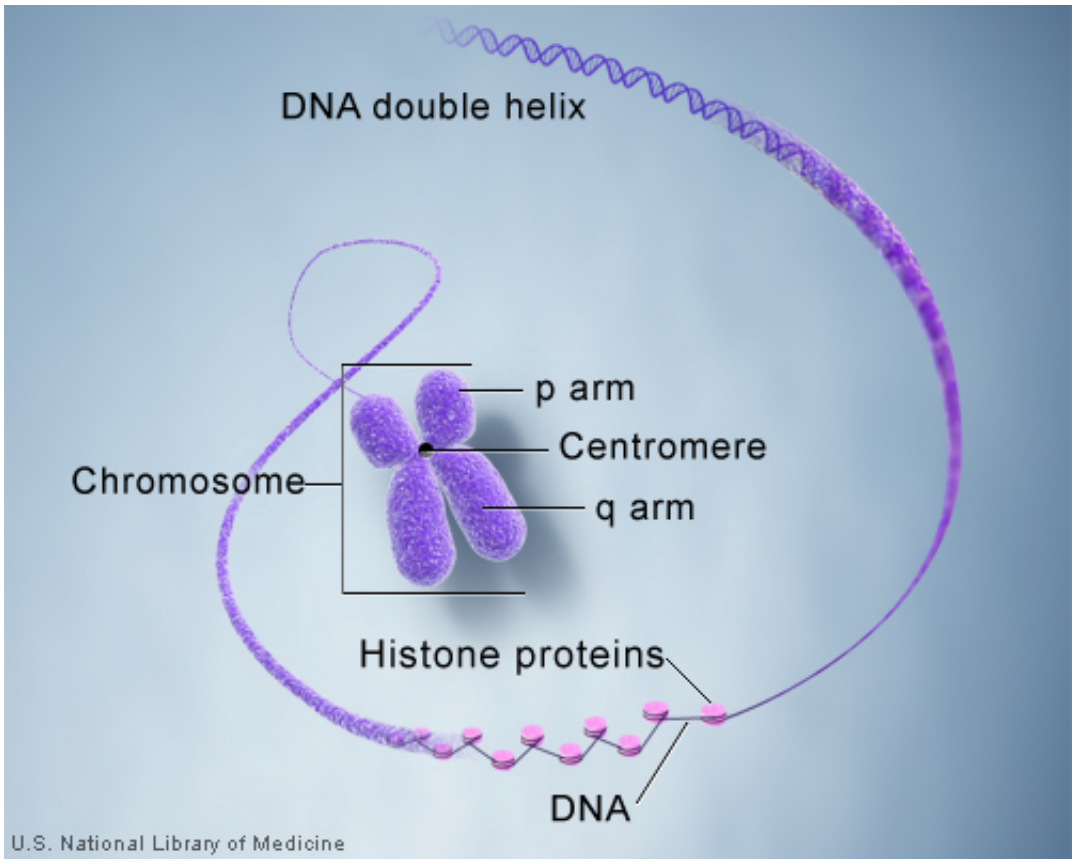
The Centre for Genetics Education offers a fact sheet that introduces genes and chromosomes (http://www.genetics.edu.au/Information/Genetics-Fact-Sheets/Genes-and-Chromosomes-FS1).

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.

For more information about chromosomes:

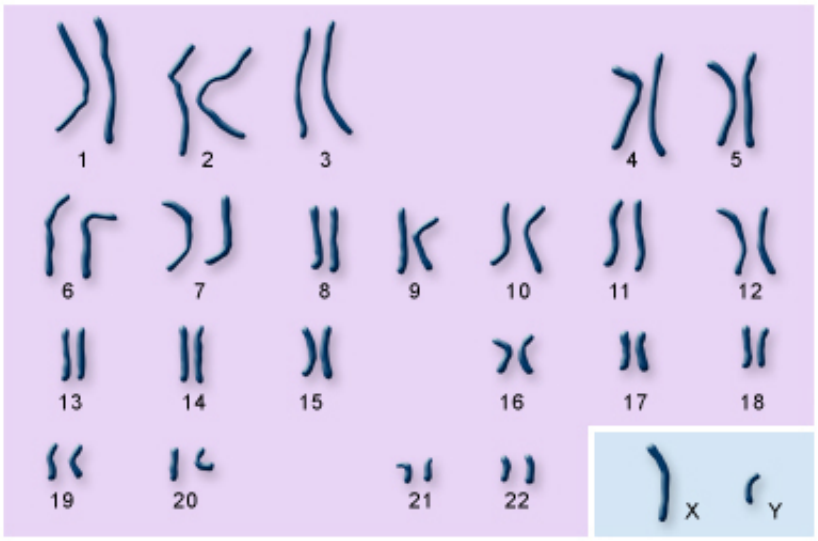
Genetics Home Reference provides information about each human chromosome (http://ghr.nlm.nih.gov/chromosomes) written in lay language.

The Centre for Genetics Education offers a fact sheet that introduces genes and chromosomes (http://www.genetics.edu.au/Information/Genetics-Fact-Sheets/Genes-and-Chromosomes-FS1).

GeneEd also provides information about the basics of chromosomes (http://geneed.nlm.nih.gov/topic_subtopic.php?tid=15&sid=17).

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.

For more information about the 23 pairs of human chromosomes:

Genetics Home Reference provides information about each human chromosome (http://ghr.nlm.nih.gov/chromosomes) written in lay language.

Chapter 2

How Genes Work

Table of Contents

What are proteins and what do they do?

Proteins are large, complex molecules that play many critical roles in the body. They do most of the work in cells and are required for the structure, function, and regulation of the body's tissues and organs.

Proteins are made up of hundreds or thousands of smaller units called amino acids, which are attached to one another in long chains. There are 20 different types of amino acids that can be combined to make a protein. The sequence of amino acids determines each protein's unique 3-dimensional structure and its specific function.

Proteins can be described according to their large range of functions in the body, listed in alphabetical order:

Examples of protein functions

Function	Description	Example
Antibody	Antibodies bind to specific foreign particles, such as viruses and bacteria, to help protect the body.	Immunoglobulin G (IgG) (illustration on page 18)
Enzyme	Enzymes carry out almost all of the thousands of chemical reactions that take place in cells. They also assist with the formation of new molecules by reading the genetic information stored in DNA.	Phenylalanine hydroxylase (illustration on page 19)
Messenger	Messenger proteins, such as some types of hormones, transmit signals to coordinate biological processes between different cells, tissues, and organs.	Growth hormone (illustration on page 20)
Structural component	These proteins provide structure and support for cells. On a larger scale, they also allow the body to move.	Actin (illustration on page 21)
Transport/storage	These proteins bind and carry atoms and small molecules within cells and throughout the body.	Ferritin (illustration on page 22)

For more information about proteins and their functions:

KidsHealth from Nemours offers a basic overview of proteins (http://kidshealth.org/kid/stay_healthy/body/protein.html) and what they do.

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