



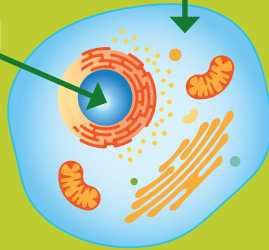
This is YOU

Your body is made up of trillions of cells

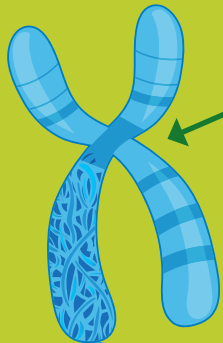
This is a cell

This is a **nucleus**

The nucleus of each cell contains **chromosomes**



This is a **chromosome**



A chromosome is made up of strands of **DNA**

This is a **DNA strand**

A section of DNA is a **gene**

Genes carry **instructions** that make you **YOU!**

All, for your one.



Learn more about genetics from these websites:

U.S. National Library of Medicine
Genetics Home Reference
ghr.nlm.nih.gov/primer

Centers for Disease Control and Prevention –
Genetics Basics
cdc.gov/genomics/about/basics.htm

Genetic Alliance
geneticalliance.org

Learn.Genetics
learn.genetics.utah.edu/content/basics

Contact Us

Lurie Children's Division of Genetics,
Genomics and Metabolism
312.227.6120

Ann & Robert H. Lurie
Children's Hospital of Chicago

225 East Chicago Avenue
Chicago, Illinois 60611-2991
312.227.4000

luriechildrens.org



The Basics of Genetics

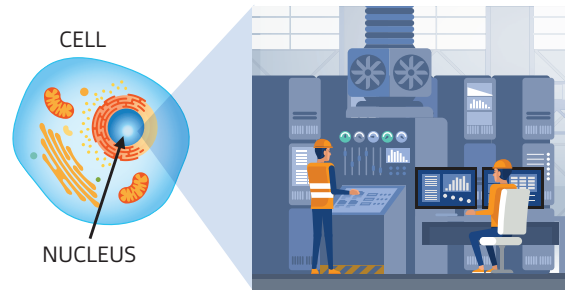
 Ann & Robert H. Lurie
Children's Hospital of Chicago®

KD.2672.06.2022

Cells

Trillions of cells make up our bodies. Each cell is like a tiny factory.

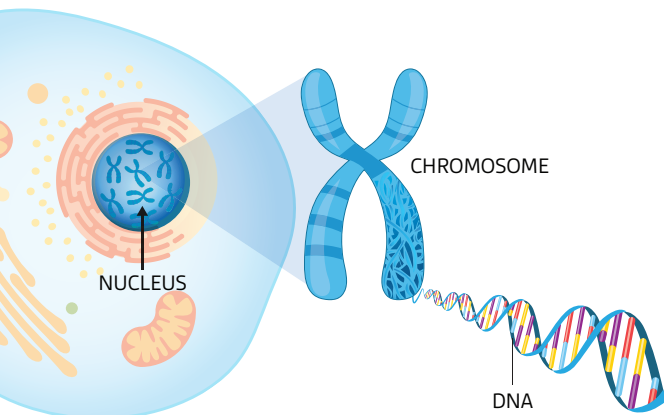
- Cells make the products our bodies need to grow and stay healthy.
- The instructions for making these products are stored in the nucleus of the cell. The nucleus is like the Control Room in a factory.



DNA, Genes and Chromosomes

A gene is a set of DNA instructions for making one of the products needed by the cell.

- There are about 20,000 genes in the human body, each with a specific job in the cell.
- Each gene is a very long string of DNA. To fit into the nucleus, the genes are tightly packaged into structures called chromosomes.
- Humans have 23 pairs of chromosomes. We receive one chromosome in each pair from our mother, and the other from our father.



Genetic Changes

The DNA instructions for our genes may vary from person to person.

- Many changes in our genes do not cause health problems. They simply make us different and unique.



For example, the genes for brown eyes and blue eyes are not exactly the same. This genetic change does not affect the ability to see.

- Some genetic changes do cause health problems and these are what we refer to as genetic conditions.
- In this case, the cell makes a product from the gene that does not work like it should. The product may be the wrong size or shape, or the cell may not make the product at all.

Let's say the cell is a factory that makes basketballs. The cell has a genetic change that causes the factory to make egg-shaped balls instead of the usual round shaped balls. The egg-shaped balls do not bounce like round balls, and players cannot use them.



- A genetic condition can come from a change in one gene, or a group of genes and/or chromosomes.

Genetic Testing

Genetic testing is performed to identify the specific genetic change that is causing a person's health problems. Genetic test results can be complicated. A member of your healthcare team who ordered the genetic test will discuss the results and answer your questions. You may also be referred to a genetic counselor, if one is not already a member of your healthcare team.

There are several types of genetic test results:

- **Positive/Abnormal** — A genetic change was identified that is known to cause a disorder that fits the person's symptoms.
- **Negative/Normal** — No genetic change that is known to cause a disorder was identified.
- **Variant of Uncertain Significance (VUS)** — A genetic change was identified, but it is unclear if this change is the cause of a person's health problems.

Limits of Genetic Testing

Genetic medicine is advancing quickly, but there are limits to what we currently know.

- Even when a genetic test comes back negative/normal, the person's health problems could still be due to a genetic change that we have not yet identified.
- A genetic diagnosis does not mean a cure is available, but it may help us:
 - Manage the symptoms and progression of the disorder.
 - Understand the natural history of the condition and what we may expect.
- Research is ongoing and there may be better treatments in the future.

Precision Medicine

Precision medicine is a new approach to treating health problems that takes into account a person's unique set of genes and environment. The goal is to match each patient with the treatment that will work best for them.