23andMe
EDUCATION

Genetics 101

A brief booklet about genetics

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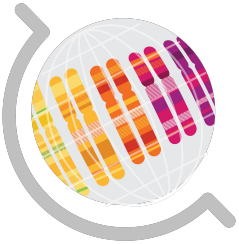

Welcome to the World of Genetics! There are so many brilliant treasures that humans have unlocked and so many mysteries that we have yet to discover in our quest to understand who we are and how we are connected to the world.

Genetics is such a fascinating topic because it's about You. Us. Our World.

It's a topic that can bring people together - people from different fields of knowledge, cultures, ethnic backgrounds, geographic locations, educational backgrounds, and more!

We all have something in common and it's inside of us.

This brief booklet will show you some of the highlights in genetics. It's an invitation for you to continue to explore, share what you've learned with your friends and families, and invite them to join you in your journey of discovery. Come, let us embark on this adventure together!

Thao Do, PhD
23andMe, Inc. Education and Academia Manager

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What is your body made of?

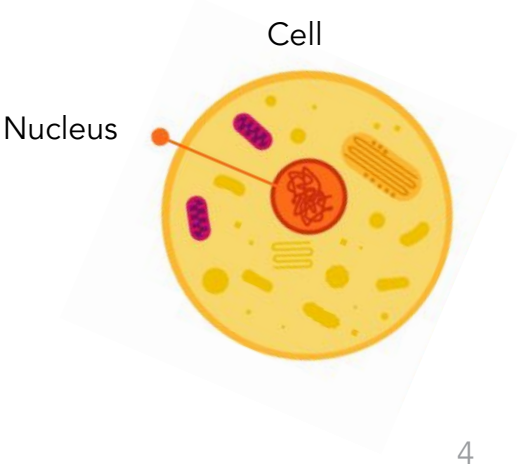
All living things are made of cells.

Your body is made of trillions of small building blocks called **cells**, like the bricks that are layered to build a house.

Cells provide the **structure** and **function** for your body.

There are many types of cells: brain cells, heart cells, muscle cells, liver cells, and more!

Almost every cell in your body has a small pouch inside called the **nucleus**. In many cells, the nucleus looks similar to an egg yolk inside an egg.



What are chromosomes?

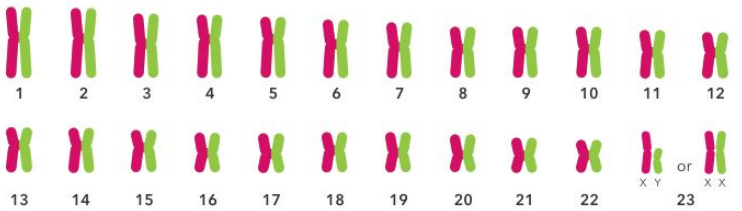
A nucleus typically contains the instructions for how cells develop and what they do. These instructions are "written" in thin strands called **chromosomes**.

Sometimes chromosomes are stretched out to be "read" and sometimes they are rolled up to be stored neatly inside the nucleus, like paper scrolls.

You inherited one set of 23 chromosomes from your dad and one set of 23 chromosomes from your mom — for a total of **23 pairs of chromosomes.**

Other living things have different numbers of chromosomes — chimpanzees have 24 pairs, bananas have 11 pairs, and fruit flies only have 4 pairs.

The number of chromosomes does not determine how complex a living thing is.



What is DNA?

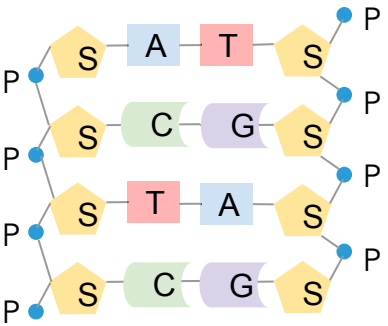
Chromosomes are made of **deoxyribonucleic acid (DNA)**. DNA is shaped like a long twisted ladder, and is often called a **double helix**.

The two sides of the ladder are made of alternating **sugar** (S) and **phosphate** (P) molecules. Each rung of the ladder is made of two **bases** connected to the sugar molecules on the sides. There are four types of bases:

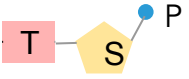
adenine (A) connects with **thymine (T)**
guanine (G) connects with **cytosine (C)**

The combination of a phosphate, a sugar, and a base is called a **nucleotide**. These basic building blocks provide the code for all living things, from roses to whales to humans — amazing!

Double helix



DNA Structure



Nucleotide

What are genes?

A **gene** is a section of DNA that contains the instructions for making materials like **proteins**.

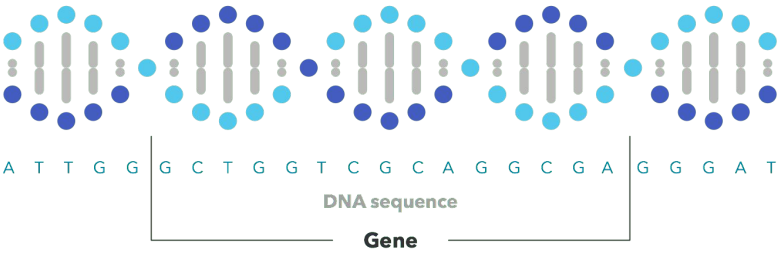
Genes are passed down from parents to children. You inherited two versions, or **alleles**, of each gene: one from mom and one from dad.

Genes are written as strings of four possible letters: A, T, C, and G, which represent the four bases.

The four bases are ordered in different ways to form thousands of genes, just like the 26 letters in the English alphabet are ordered in different ways to form thousands of words.

All of your genetic material combined, including your genes, make up your **genome.**

Genetics is the study of genes.



How do genes influence your cells' growth and function?

All of your cells have the same genes. However, each cell turns on, or **expresses**, only a portion of the genes to produce the proteins that your body needs at that moment in time. The cell turns off, or **not expresses**, the rest of the genes.

Genes can be turned on or off at different times in your life, like light switches, to allow your body to grow and to quickly react to environmental changes.

The process of turning genes on and off is called **gene regulation**.

Your body uses gene regulation to determine which cells to make, like brain cells in your brain and liver cells in your liver.

Your body also uses gene regulation to determine when to make certain cells, like rapidly make lots of white blood cells when you are sick from an infection and slow down production when you have recovered.

What are proteins?

Your genes provide the instructions to make proteins.

Each protein is made of a chain of molecules called amino acids that are linked together, like a beaded necklace is made of a string of colorful beads.

There are 20 different amino acids.

Based on the instructions coded in your genes, each amino acid is added one-by-one to make a protein.

Proteins are molecules that can move and change shapes to do different jobs.

Some proteins provide the structure for your cells, similar to the way your skeleton supports your body.

Other proteins break down food, transport oxygen, and detect invading bacteria and viruses.

Quick summary

Your body is made of cells.

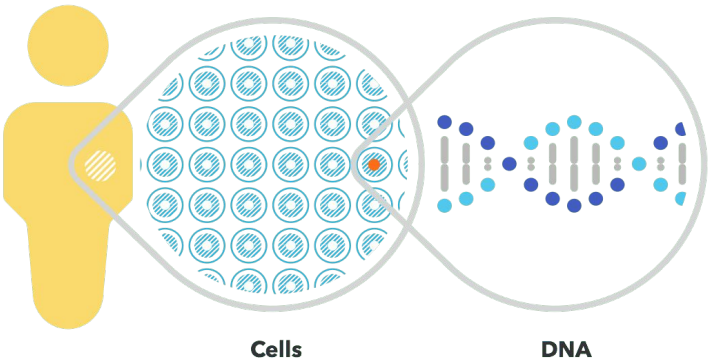
Almost all of your cells contain a nucleus.

Each nucleus contains 23 pairs of chromosomes.

Chromosomes are made of DNA.

Genes are sections of DNA that contain the instructions for making materials like proteins.

Proteins do important jobs in your body like providing the structure and function for your cells.



What is the difference between genotype and phenotype?

Your **genotype** is your unique set of genes. Your genes, lifestyle, and environment help determine different characteristics, like your traits, ancestry, and health conditions. These characteristics are called **phenotype**.

What is the difference between sequencing and genotyping?

Sequencing and **genotyping** are two different laboratory processes for analyzing DNA.

Sequencing determines the exact sequence of a certain length of DNA. Genotyping determines which genetic variants of interest an individual possesses.

So, sequencing is like listening to all of a singer's songs in order, and genotyping is like listening to just the singer's greatest hits.

Sequencing Genotyping

What are variants? What are SNPs?

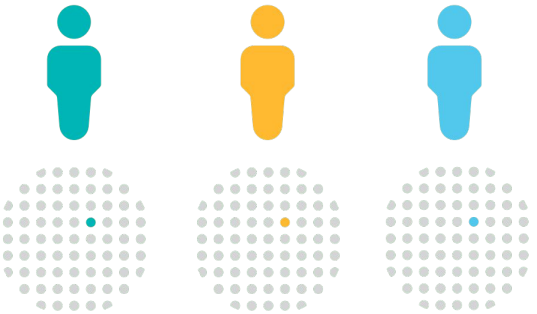
All humans have the same set of genes.

However, there are small genetic differences between people; these differences are called **variants**.

There are many types of genetic variations. The most common type of genetic variation is a **single nucleotide polymorphism (SNP; pronounced "snip")** — 'poly' means 'many' and 'morphism' means 'form.' A SNP is a difference in one nucleotide at a specific location in the genome.

For example, at the same location in a gene that codes for hair color, one person might have a nucleotide with a base A, while another person has a nucleotide with a base C. In this case, there are two different versions of the hair color gene.

Many genetic variants do not have any effect, but some are associated with certain characteristics like traits, ancestry, and health conditions.



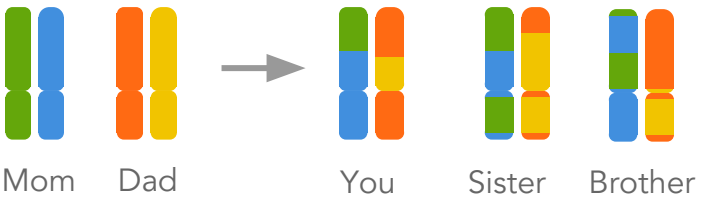
What is genetic recombination?

Several biological processes exist to increase **genetic diversity**. One process is **genetic recombination**, which is when your mom's and dad's DNA gets randomly shuffled before being passed on to you.

For example, in this image, your mom has a green chromosome that she inherited from her mom (your grandmother) and a blue chromosome that she inherited from her dad (your grandfather).

Your mom does not pass either a green or blue chromosome to you. Instead, she passes a random mixture of her two chromosomes - a blue/green chromosome - to you. Similarly, your dad passes a random mixture of his two chromosomes - an orange/yellow chromosome - to you.

This random shuffling of DNA makes you genetically unique. There is one exception: if you have an **identical twin**, both of you would have the exact same genes.



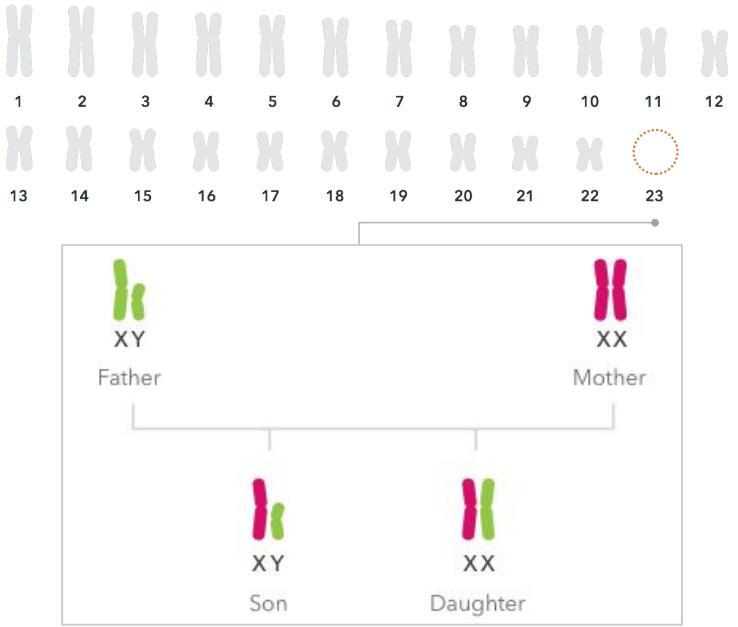
How does DNA influence your genetic sex?

The 23rd chromosome pair contains the **sex chromosomes**, which determines whether you are genetically male or female.

In most cases, regardless of your genetic sex, your mom passes one X chromosome to you.

If your dad passes one Y chromosome to you, then you are genetically male with XY chromosomes.

If your dad passes one X chromosome to you, then you are genetically female with XX chromosomes.



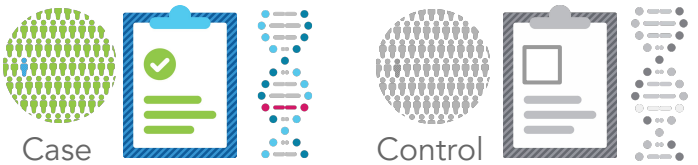
What is GWAS?

Researchers analyze millions of individuals' SNPs (genotype) to understand how genetics influences traits, ancestry, and health (phenotype).

For example, in a **research survey,** scientists may ask thousands of people who have consented to research "Have you been diagnosed with a condition?" Some answer "Yes" - these participants are added in the **case group**. Some answer "No" - these participants are added in the **control group**.

Scientists conduct experiments called **Genome-Wide Association Studies (GWAS)** to analyze millions of SNPs in individuals in both the case and control groups.

Sometimes, scientists observe that specific genotypes are often found in people with specific phenotypes - these are called **genetic associations**. These insights can help scientists understand how our genetics influence our characteristics and develop new preventions and treatments for medical conditions.



Is genetics the only factor that determines your traits?

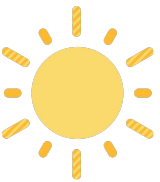
Genetics can tell you a lot about yourself, but it is *not* the only factor. **Your genetics, lifestyle, and environment can influence who you are.**

For example, just about everybody is born with the ability to digest lactose, and this ability allows babies to live and grow by drinking their mothers' breast milk. As children grow older and begin to eat different foods, many of them lose the ability to produce the lactase enzyme to digest the sugar lactose found in dairy products like milk, cheese, and yogurt. These adults may experience bloating, diarrhea and gas after eating or drinking dairy products - a condition known as **lactose intolerance**.

Research suggests that ancient humans were lactose intolerant. In some geographic regions, as humans started domesticating cows, goats, and sheeps, and heavily depended on their milk for nutrients, new genetic variants appeared that gave some groups of people the ability to digest lactose in adulthood.

Genetics

Lifestyle



Environment

There is still a lot more to learn!

As scientists collect and analyze more data, we will learn more about how genetics influences who we are, how we are connected to each other, how we can prevent and treat human diseases, and how we can live healthy lives.

Understanding our genome is one of the most exciting scientific discoveries of our lifetime.

Now *you* have the opportunity to learn more about genetics and share your knowledge with your community!

Special thanks

Special thanks to contributing writers, reviewers, and editors: Thao Do, Sarah Laskey, Becca Krock, Ruth Tennen, Hoang Nhan, Anjali Shastri, James Ashenhurst, Chelsea Robertson, Kayla Capper, Alison Chubb, Mark Pisaro, Nicole Faria, Lisa Charter, Stacey Detweiler, Jeffrey Pollard, and Raniele Jago.

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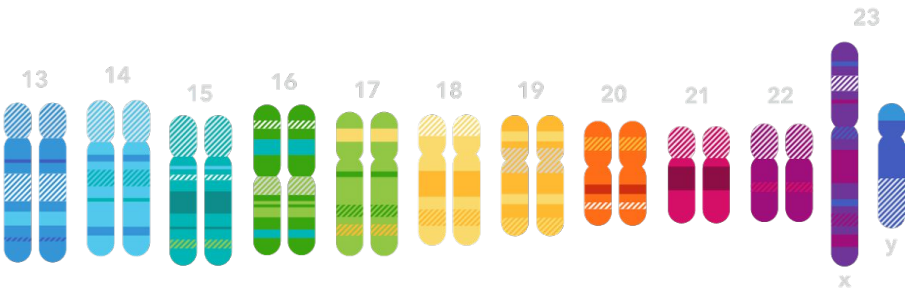

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