Chromosomes

DNA, chromosomes, and genomes. Homologous chromosomes, sister chromatids, and haploid/diploid.

Introduction

When a cell divides, one of its main jobs is to make sure that each of the two new cells gets a full, perfect copy of genetic material. Mistakes during copying, or unequal division of the genetic material between cells, can lead to cells that are unhealthy or dysfunctional (and may lead to diseases such as cancer).

But what exactly is this genetic material, and how does it behave over the course of a cell division?

DNA and genomes

DNA (**deoxyribonucleic acid**) is the genetic material of living organisms. In humans, DNA is found in almost all the cells of the body and provides the instructions they need to grow, function, and respond to their environment.

When a cell in the body divides, it will pass on a copy of its DNA to each of its daughter cells. DNA is also passed on at the level of organisms, with the DNA in sperm and egg cells combining to form a new organism that has genetic material from both its parents.

Physically speaking, DNA is a long string of paired chemical units (nucleotides) that come in four different types, abbreviated A, T, C, and G, and it carries information organized into units called **genes**. Genes typically provide instructions for making proteins, which give cells and organisms their functional characteristics.

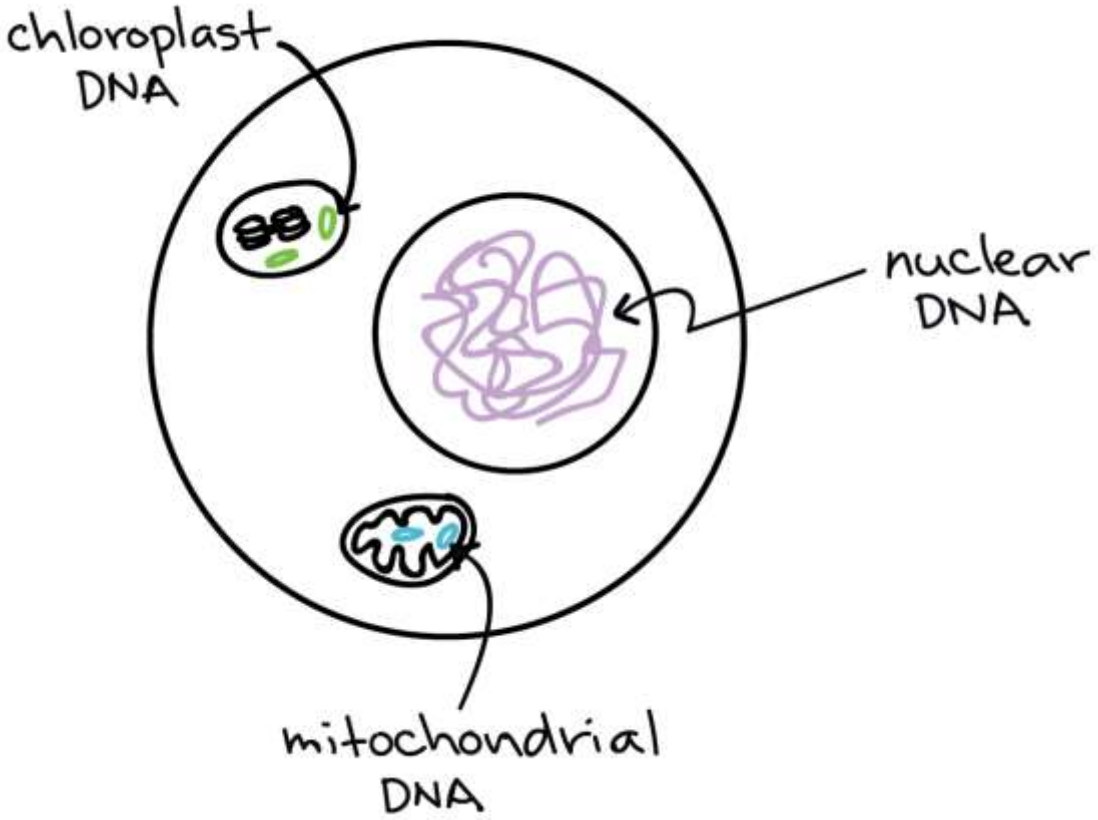


Image of a eukaryotic cell, showing the nuclear DNA (in the nucleus), the mitochondrial DNA (in the mitochondrial matrix, and the chloroplast DNA (in the stroma of the chloroplast.

In eukaryotes such as plants and animals, the majority of DNA is found in the nucleus and is called **nuclear DNA**. Mitochondria, organelles that harvest energy for the cell, contain their own **mitochondrial DNA**, and chloroplasts, organelles that carry out photosynthesis in plant cells, also have **chloroplast DNA**. The amounts of DNA found in mitochondria and chloroplasts are much smaller than the amount found in the nucleus. In bacteria, most of the DNA is found in a central region of the cell called the **nucleoid**, which functions similarly to a nucleus but is not surrounded by a membrane.

A cell's set of DNA is called its **genome**. Since all of the cells in an organism (with a few exceptions) contain the same DNA, you can also say that an organism has its own genome, and since the members of a species typically have similar genomes, you can also describe the genome of a species. In general, when people refer to the human genome, or any other eukaryotic genome, they mean the set of DNA found in the nucleus. Mitochondria and chloroplasts are considered to have their own separate genomes.

Chromatin

In a cell, DNA does not usually exist by itself, but instead associates with specialized proteins that organize it and give it structure. In eukaryotes, these proteins include the **histones**, a group of basic (positively charged) proteins that form "bobbins" around which negatively charged DNA can wrap. In addition to organizing DNA and making it more compact, histones play an important role in determining which genes are active. The complex of DNA plus histones and other structural proteins is called **chromatin**.

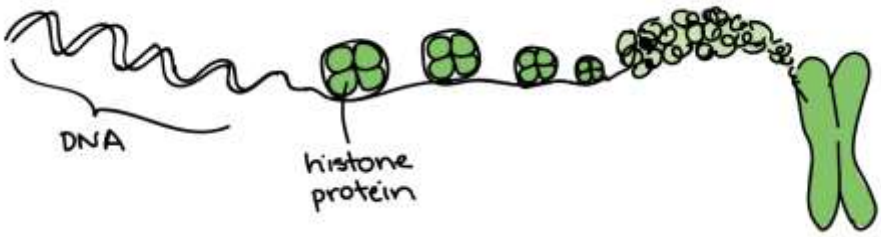


Image of a long, double-stranded DNA polymer, which wraps around clusters of histone proteins. The DNA wrapped around histones is further organized into higher-order structures that give a chromosome its shape.

For most of the life of the cell, chromatin is **decondensed**, meaning that it exists in long, thin strings that look like squiggles under the microscope. In this state, the DNA can be accessed relatively easily by cellular machinery (such as proteins that read and copy DNA), which is important in allowing the cell to grow and function.

Decondensed may seem like an odd term for this state – why not just call it "stringy"? – but makes more sense when you learn that chromatin can also **condense**. Condensation takes place when the cell is about to divide. When chromatin condenses, you can se that eukaryotic DNA is not just one long string. Instead, it's broken up into separate, linear pieces called **chromosomes**. Bacteria also have chromosomes, but their chromosomes are typically circular.

Chromosomes

Each species has its own characteristic number of chromosomes. Humans, for instance, have 46 chromosomes in a typical body cell (somatic cell), while dogs have 78. Like many species of animals and plants, humans are **diploid** (**2*n***), meaning that most of their chromosomes come in matched sets known as **homologous pairs**. The 46 chromosomes of a human cell are organized into 23 pairs, and the two members of each pair are said to be **homologues** of one another (with the slight exception of the X and Y chromosomes; see below).

Human sperm and eggs, which have only one homologous chromosome from each pair, are said to be **haploid** (**1*n***). When a sperm and egg fuse, their genetic material combines to form one complete, diploid set of chromosomes. So, for each homologous pair of chromosomes in your genome, one of the homologues comes from your mom and the other from your dad.

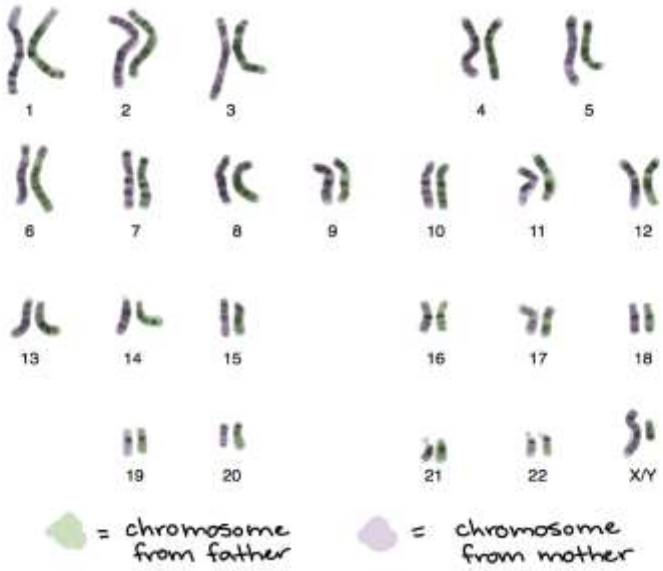


Image of the karyotype of a human male, with chromosomes from the mother and father false-coloured purple and green, respectively.

Image modified from "Karyotype," by the National Institutes of Health (public domain).
The two chromosomes in a homologous pair are very similar to one another and have the same size and shape. Most importantly, they carry the same type of genetic information: that is, they have the same genes in the same locations. However, they don't necessarily have the same versions of genes. That's because you may have inherited two different gene versions from your mom and your dad.

As a real example, let's consider a gene on chromosome 9 that determines blood type (A, B, AB, or O). It's possible for a person to have two identical copies of this gene, one on each homologous chromosome—for example, you may have a double dose of the gene version for type A. On the other hand, you may have two different gene versions on your two homologous chromosomes, such as one for type A and one for type B (giving AB blood).

The **sex chromosomes**, X and Y, determine a person's biological sex: XX specifies female and XY specifies male. These chromosomes are not true homologues and are an exception to the rule of the same genes in the same places. Aside from small regions of similarity needed during meiosis, or sex cell production, the X and Y chromosomes are different and carry different genes. The 44 non-sex chromosomes in humans are called **autosomes**.

Chromosomes and cell division

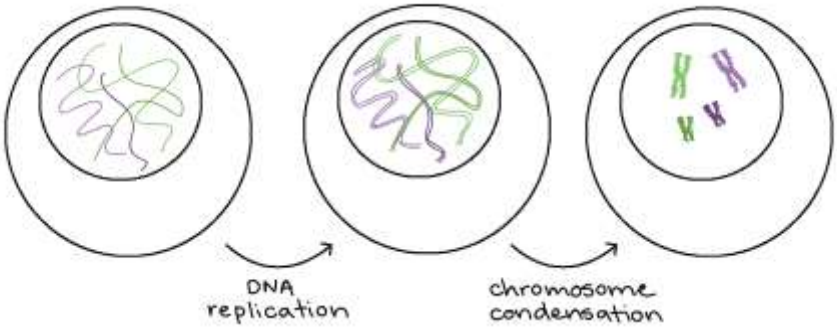
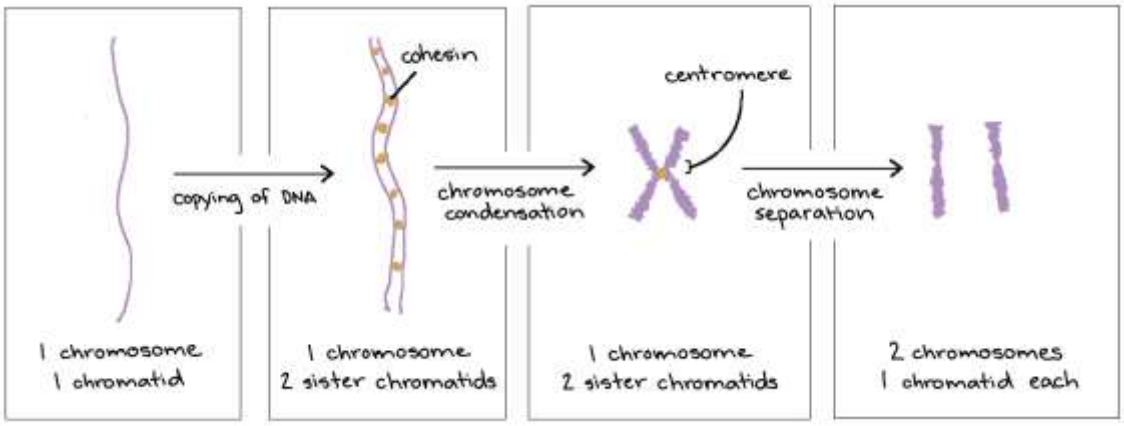


Image of a cell undergoing DNA replication (all the chromosomes in the nucleus are copied) and chromosome condensation (all the chromosomes become compact). In the first image, there are four decondensed, stringy chromosomes in the nucleus of the cell. After DNA replication, each chromosome now consists of two physically attached sister chromatids. After chromosome condensation, the chromosomes condense to form compact structures (still made up of two chromatids).

As a cell prepares to divide, it must make a copy of each of its chromosomes. The two copies of a chromosome are called **sister chromatids**. The sister chromatids are identical to one another and are attached to each other by proteins called **cohesins**. The attachment between sister chromatids is tightest at the **centromere**, a region of DNA that is important for their separation during later stages of cell division.

As long as the sister chromatids are connected at the centromere, they are still considered to be one chromosome. However, as soon as they are pulled apart during cell division, each is considered a separate chromosome.



What happens to a chromosome as a cell prepares to divide.

1. The chromosome consists of a single chromatid and is decondensed (long and string-like).
2. The DNA is copied. The chromosome now consists of two sister chromatids, which are connected by proteins called cohesins.
3. The chromosome condenses. It is still made up of two sister chromatids, but they are now short and compact rather than long and stringy. They are most tightly connected at the centromere region, which is the inward-pinching "waist" of the chromosome.
4. The chromatids are pulled apart. Each is now considered its own chromosome.

Why do cells put their chromosomes through this process of replication, condensation, and separation? The short answer is: to make sure that, during cell division, each new cell gets exactly one copy of each chromosome.

For a more satisfying answer, check out the articles and videos on the cell cycle and mitosis. There, you can see how the behaviour of chromosomes helps cells pass on a perfect set of DNA to each daughter cell during division.