

Human Chromosomes and Genes

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CHAPTER

1

Human Chromosomes and Genes

- Define chromosome.
- Describe human autosomes and sex chromosomes.
- Summarize the importance of alleles in humans.



Coiled bundles of DNA and proteins, containing hundreds or thousands of genes. What are these things?

Chromosomes. These ensure that each cell receives the proper amount of DNA during cell division. And usually people have 46 of them, 23 from each parent.

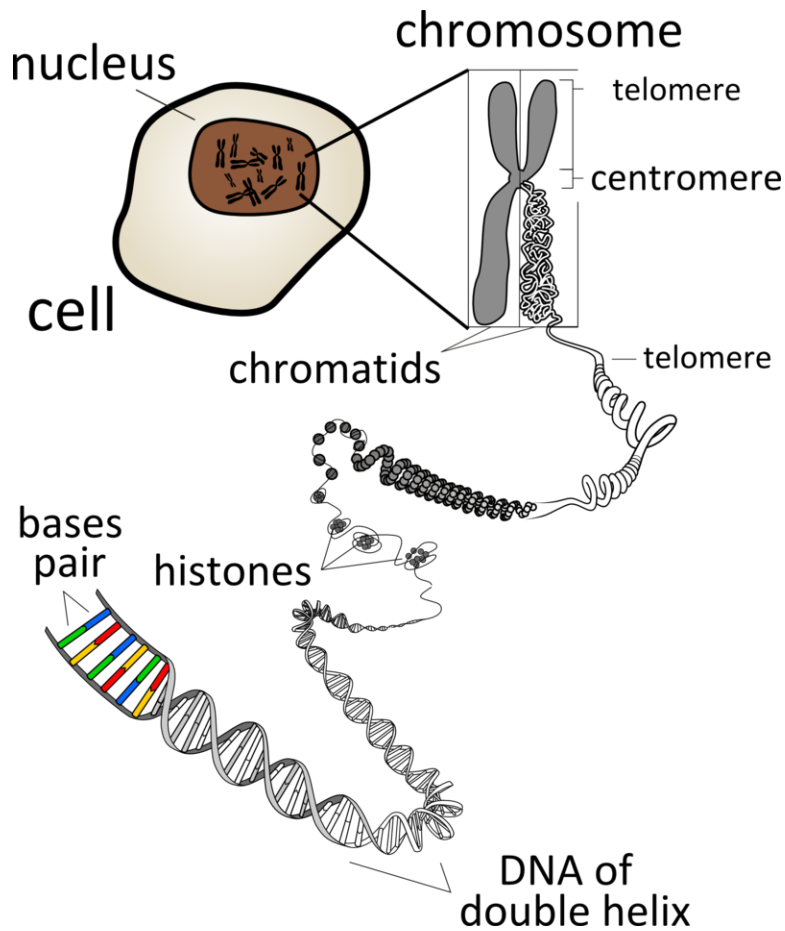
Chromosomes and Genes

Each species has a characteristic number of chromosomes. **Chromosomes** are coiled structures made of DNA and proteins called **histones** (**Figure 1.1**). Chromosomes are the form of the genetic material of a cell during cell division. See the "Chromosomes" section for additional information.

The human species is characterized by 23 pairs of chromosomes, as shown in **Figure 1.2**. You can watch a short animation about human chromosomes at this link: <http://www.dnalc.org/view/15520-DNA-is-organized-into-46-chromosomes-including-sex-chromosomes-3D-animation.html> .

Autosomes

Of the 23 pairs of human chromosomes, 22 pairs are autosomes (numbers 1–22 in **Figure 1.2**). **Autosomes** are chromosomes that contain genes for characteristics that are unrelated to sex. These chromosomes are the same in males and females. The great majority of human genes are located on autosomes. At the link below, you can click on any human chromosome to see which traits its genes control. http://www.ornl.gov/sci/techresources/Human_Genome/posters/chromosome/chooser.shtml

**FIGURE 1.1**

The human genome has 23 pairs of chromosomes located in the nucleus of somatic cells. Each chromosome is composed of genes and other DNA wound around histones (proteins) into a tightly coiled molecule.

Sex Chromosomes

The remaining pair of human chromosomes consists of the **sex chromosomes**, X and Y. Females have two X chromosomes, and males have one X and one Y chromosome. In females, one of the X chromosomes in each cell is inactivated and known as a Barr body. This ensures that females, like males, have only one functioning copy of the X chromosome in each cell.

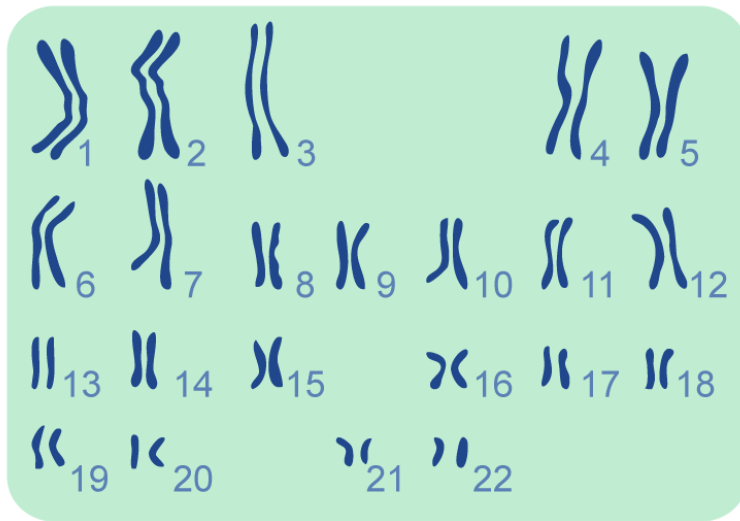
As you can see from **Figure 1.1** and **Figure 1.2**, the X chromosome is much larger than the Y chromosome. The X chromosome has about 2,000 genes, whereas the Y chromosome has fewer than 100, none of which are essential to survival. (For comparison, the smallest autosome, chromosome 22, has over 500 genes.) Virtually all of the X chromosome genes are unrelated to sex. Only the Y chromosome contains genes that determine sex. A single Y chromosome gene, called **SRY** (which stands for sex-determining region Y gene), triggers an embryo to develop into a male. Without a Y chromosome, an individual develops into a female, so you can think of female as the default sex of the human species. Can you think of a reason why the Y chromosome is so much smaller than the X chromosome? At the link that follows, you can watch an animation that explains why: http://www.hhmi.org/biointeractive/gender/Y_evolution.html .

Human Genes

Humans have an estimated 20,000 to 22,000 genes. This may sound like a lot, but it really isn't. Far simpler species have almost as many genes as humans. However, human cells use splicing and other processes to make multiple proteins from the instructions encoded in a single gene. Of the 3 billion base pairs in the human genome, only about

Human chromosomes (23 pairs)

Autosomes



Sex chromosomes

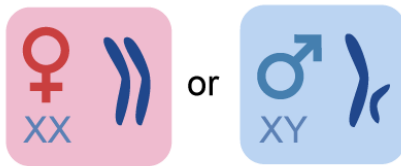


FIGURE 1.2

Human Chromosomes. Humans have 23 pairs of chromosomes. Pairs 1-22 are autosomes. Females have two X chromosomes, and males have an X and a Y chromosome.

25 percent make up genes and their regulatory elements. The functions of many of the other base pairs are still unclear. To learn more about the coding and noncoding sequences of human DNA, watch the animation at this link: http://www.hhmi.org/biointeractive/dna/DNAi_coding_sequences.html .

The majority of human genes have two or more possible **alleles**, which are alternative forms of a gene. Differences in alleles account for the considerable genetic variation among people. In fact, most human genetic variation is the result of differences in individual DNA bases within alleles.

Summary

- Humans have 23 pairs of chromosomes. Of these, 22 pairs are autosomes.
- The X and Y chromosomes are the sex chromosomes. Females have two X chromosomes, and males have one X and one Y.
- Human chromosomes contain a total of 20,000 to 22,000 genes, the majority of which have two or more alleles.

Explore More

Use this resource to answer the questions that follow.

- **Chromosomes** at <http://www.genome.gov/26524120> .

1. What is a chromosome?

2. What is the role of chromosomes during cell division?
3. Do all living things have the same types of chromosomes?
4. What are centromeres? What is their role?
5. What are telomeres? What is their role?

Review

1. Describe human chromosomes.
2. Compare and contrast human autosomes and sex chromosomes.
3. What is SRY?
4. Why are females the "default sex" of the human species?

References

1. User:KES47/Wikimedia Commons. [Zooming in on chromosome and chromatids](#) . CC BY 3.0
2. Mariana Ruiz Villarreal (LadyofHats) for CK-12 Foundation. [Karyotype, showing 23 chromosomes](#) . CC BY-NC 3.0