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Chromosome

Contributors: Tara C. Smith

Edited by: Sarah Boslaugh

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A chromosome is a large macromolecule that functions as the structural unit of the genetic material. In eukaryotes, chromosomes are molecules consisting of linear, double-stranded deoxyribonucleic acid (DNA) and associated proteins. In prokaryotes, chromosomes are typically single-stranded, circular molecules. In epidemiology, knowledge of chromosomes is essential because chromosomal abnormalities are a leading cause of human genetic diseases. These abnormalities can include deletions (where part or all of a chromosome is missing), duplications (part or all of a chromosome is duplicated, resulting in excess genetic material), translocations (part of a chromosome is transferred to another chromosome), or inversions (part of the chromosome has detached, 'flipped over,' and reattached, resulting in the genetic material being in the wrong order).

The totality of all the chromosomes in an individual is referred to as its genome. Each chromosome consists of genes (functional regions of the DNA that encode proteins), noncoding DNA, and associated structural proteins and ribonucleic acid (RNA). The sum of the material that makes up the chromosome is called chromatin. The number of chromosomes present varies greatly between species, ranging from a single chromosome (in the case of many bacteria) to more than 50 chromosomes in many animals. Humans have 46 paired chromosomes, receiving 23 from each parent. These include 22 pairs of autosomal chromosomes and one pair of sex chromosomes, which determine the gender of the individual: Females receive an X chromosome from each parent (giving them an XX genotype), while males receive an X chromosome from their mother and a Y chromosome from their father (resulting in an XY genotype).

One of the most important genetic diseases is Down's syndrome, which is the result of a trisomy (a duplication, leading to three copies) of chromosome 21. Individuals with Down's syndrome typically have mild to moderate mental retardation, decreased muscle tone, and shortened limbs. The incidence of Down's syndrome is approximately 1 in 800 births in the United States, and is most common in mothers who are above the age of 40 at birth. However, genetic testing can be carried out during pregnancy to inform parents if their fetus is positive for this chromosomal abnormality.

While Down's syndrome results from an extra autosomal chromosome, other genetic conditions can result from duplication or deletion of all or portions of the sex chromosomes (X and Y). Men with Klinefelter's syndrome possess an extra X chromosome, leading to an XXY genotype. Physically, they tend to be sterile, and tall with long arms and legs. Females with Turner syndrome, on the other hand, lack a second sex chromosome, and genetically are XO. Female sex characteristics may be present but are underdeveloped.

Chromosomal abnormalities also play a role in some cancers. Tumor cells frequently are aneuploid, meaning they have an abnormal number of chromosomes. They may also contain translocations or portions of chromosomes that have been copied not just once but dozens or hundreds of times.

Tara C. Smith

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See also

- [Genetic Disorders](#)

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Further Readings

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