A **chromosome** (from <u>ancient Greek</u>: χρωμόσωμα, *chromosoma, chroma* means color, *soma* means body) is a <u>DNA</u> molecule with part or all of the genetic material (<u>genome</u>) of an organism.

Chromosomes are normally visible under a <u>light microscope</u> only when the cell is undergoing the <u>metaphase</u> of <u>cell division</u>. Before this happens, every chromosome is copied once (<u>S</u> <u>phase</u>), and the copy is joined to the original by a <u>centromere</u>, resulting in an X-shaped structure. The original chromosome and the copy are now called <u>sister chromatids</u>. During metaphase the X-shape structure is called a metaphase chromosome. In this highly condensed form chromosomes are easiest to distinguish and study.^[11] In animal cells, chromosomes reach their highest compaction level in anaphase during segregation.^[2]

Chromosomes vary widely between different <u>organisms</u>. Some species such as certain <u>bacteria</u>, which lack <u>histones</u>, also contain <u>plasmids</u> or other <u>extrachromosomal DNA</u>. These are circular structures in the <u>cytoplasm</u> that contain cellular DNA and play a role in <u>horizontal gene</u> <u>transfer</u>.^[11] In prokaryotes (see <u>nucleoids</u>) and <u>viruses</u>,^[3] the DNA is often densely packed and organized; in the case of <u>archaea</u>, by homology to eukaryotic histones, and in the case of bacteria, by <u>histone-like</u> proteins.

<u>DNA condensation</u> of the duplicated chromosomes during <u>cell division</u> (<u>mitosis</u> or <u>meiosis</u>) results either in a four-arm structure (pictured to the right) if the <u>centromere</u> is located in the middle of the chromosome or a two-arm structure if the centromere is located near one of the ends. Chromosomal <u>recombination</u> during meiosis and subsequent <u>sexual reproduction</u> play a significant role in genetic diversity. If these structures are manipulated incorrectly, through processes known as chromosomal instability and translocation, the cell may undergo <u>mitotic catastrophe</u> and die, or it may unexpectedly evade <u>apoptosis</u>, leading to the progression of <u>cancer</u>.

Some use the term chromosome in a wider sense, to refer to the individualized portions of chromatin in cells, either visible or not under light microscopy. However, others use the concept in a narrower sense, to refer to the individualized portions of chromatin during cell division, visible under light microscopy due to high condensation.

Etymology

Emilio Battaglia (1917-2011) points out that many of the most familiar caryological terms are inadequate or illogical or, in some cases, etymologically incorrect so that they should be replaced by more adequate alternatives suggested by the present scientific progress. The author has been particularly disappointed by the illogicality of the present chromosomal (chromatin-chromosome) terminology based on, or inferred by, two terms, Chromatin (Flemming 1880) and Chromosom (Waldeyer 1888), both inappropriately ascribed to a basically non coloured state.

History of discovery



<u>Walter Sutton</u> (left) and <u>Theodor Boveri</u> (right) independently developed the chromosome theory of inheritance in 1902.

The word *chromosome* (/'kroumə soum, - zoum/ comes from the <u>Greek</u> $\chi \rho \tilde{\omega} \mu \alpha$ (*chroma*, "colour") and $\sigma \tilde{\omega} \mu \alpha$ (*soma*, "body"), describing their strong staining by particular <u>dyes</u>.

<u>Schleiden</u>,^[1] <u>Virchow</u> and <u>Bütschli</u> were among the first scientists who recognized the structures now familiar as chromosomes. The term was coined by <u>von Waldeyer-Hartz</u>, referring to the term <u>chromatin</u>, which was introduced by <u>Walther Flemming</u>.

In a series of experiments beginning in the mid-1880s, <u>Theodor Boveri</u> gave the definitive demonstration that chromosomes are the <u>vectors</u> of <u>heredity</u>. His two principles were the *continuity* of chromosomes and the *individuality* of chromosomes. It is the second of these principles that was so original.<u>Wilhelm Roux</u> suggested that each chromosome carries a different <u>genetic load</u>. Boveri was able to test and confirm this hypothesis. Aided by the rediscovery at the start of the 1900s of <u>Gregor Mendel</u>'s earlier work, Boveri was able to point out the connection between the rules of inheritance and the behaviour of the chromosomes. Boveri influenced two generations of American cytologists: <u>Edmund Beecher Wilson</u>, <u>Nettie</u> <u>Stevens</u>, <u>Walter Sutton</u> and <u>Theophilus Painter</u> were all influenced by Boveri (Wilson, Stevens, and Painter actually worked with him).

In his famous textbook *The Cell in Development and Heredity*, Wilson linked together the independent work of Boveri and Sutton (both around 1902) by naming the chromosome theory of inheritance the <u>Boveri–Sutton chromosome theory</u> (the names are sometimes reversed). <u>Ernst</u> <u>Mayr</u> remarks that the theory was hotly contested by some famous geneticists: <u>William</u> <u>Bateson</u>, <u>Wilhelm Johannsen</u>, <u>Richard Goldschmidt</u> and <u>T.H. Morgan</u>, all of a rather dogmatic turn of mind. Eventually, complete proof came from chromosome maps in Morgan's own lab.

The number of human chromosomes was published in 1923 by <u>Theophilus Painter</u>. By inspection through the microscope, he counted 24 pairs, which would mean 48 chromosomes. His error was copied by others and it was not until 1956 that the true number, 46, was determined by Indonesia-born cytogeneticist <u>Joe Hin Tjio</u>.

Prokaryotes

The prokaryotes – <u>bacteria</u> and <u>archaea</u> – typically have a single <u>circular chromosome</u>, but many variations exist. The chromosomes of most bacteria, which some authors prefer to call <u>genophores</u>, can range in size from only 130,000 <u>base pairs</u> in the <u>endosymbiotic</u> bacteria <u>Candidatus Hodgkinia cicadicola^[17]</u> and <u>Candidatus Tremblaya</u> <u>princeps</u>, to more than 14,000,000 base pairs in the soil-dwelling bacterium <u>Sorangium</u> <u>cellulosum</u>. Spirochaetes of the <u>genus</u> *Borrelia* are a notable exception to this arrangement, with bacteria such as <u>Borrelia burgdorferi</u>, the cause of Lyme disease, containing a single *linear* chromosome.

Structure in sequences.

Prokaryotic chromosomes have less sequence-based structure than eukaryotes. Bacteria typically have a one-point (the <u>origin of replication</u>) from which replication starts, whereas some archaea contain multiple replication origins. The genes in prokaryotes are often organized in <u>operons</u>, and do not usually contain <u>introns</u>, unlike eukaryotes.

DNA packaging

<u>Prokaryotes</u> do not possess nuclei. Instead, their DNA is organized into a structure called the <u>nucleoid</u>. The nucleoid is a distinct structure and occupies a defined region of the bacterial cell. This structure is, however, dynamic and is maintained and remodeled by the actions of a range of histone-like proteins, which associate with the bacterial chromosome. In <u>archaea</u>, the DNA in chromosomes is even more organized, with the DNA packaged within structures similar to eukaryotic nucleosomes. Bacterial chromosomes tend to be tethered to the <u>plasma</u> <u>membrane</u> of the bacteria. In molecular biology application, this allows for its isolation from plasmid DNA by centrifugation of lysed bacteria and pelleting of the membranes (and the attached DNA).

Prokaryotic chromosomes and plasmids are, like eukaryotic DNA, generally <u>supercoiled</u>. The DNA must first be released into its relaxed state for access for <u>transcription</u>, regulation, and <u>replication</u>.