

Genome

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In modern molecular biology and genetics, a **genome** is the **genetic material** of an organism. It consists of DNA (or RNA in RNA viruses). The genome includes both the genes, (the coding regions), the noncoding DNA^[1] and the genomes of the mitochondria^[2] and chloroplasts.

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An image of the 46 chromosomes making up the diploid genome of a human male. (The mitochondrial chromosome is not shown.)

Origin of term

The term *genome* was created in 1920 by Hans Winkler,^[3] professor of botany at the University of Hamburg, Germany. The Oxford Dictionary suggests the name is a blend of the words *gene* and *chromosome*.^[4] However, see omics for a more thorough discussion. A few related *-ome* words already existed—such as *biome*, *rhizome*, forming a vocabulary into which *genome* fits systematically.^[5]

Overview

Some organisms have multiple copies of chromosomes: diploid, triploid, tetraploid and so on. In classical genetics, in a sexually reproducing organism (typically eukarya) the gamete has half the number of chromosomes of the somatic cell and the genome is a full set of chromosomes in a diploid

cell. The halving of the genetic material in gametes is accomplished by the segregation of homologous chromosomes during meiosis.^[6] In haploid organisms, including cells of bacteria, archaea, and in organelles including mitochondria and chloroplasts, or viruses, that similarly contain genes, the single or set of circular or linear chains of DNA (or RNA for some viruses), likewise constitute the genome. The term *genome* can be applied specifically to mean what is stored on a complete set of nuclear DNA (i.e., the "nuclear genome") but can also be applied to what is stored within organelles that contain their own DNA, as with the "mitochondrial genome" or the "chloroplast genome". Additionally, the genome can comprise non-chromosomal genetic elements such as viruses, plasmids, and transposable elements.^[7]

Typically, when it is said that the genome of a sexually reproducing species has been "sequenced", it refers to a determination of the sequences of one set of autosomes and one of each type of sex chromosome, which together represent both of the possible sexes. Even in species that exist in only one sex, what is described as a "genome sequence" may be a composite read from the chromosomes of various individuals. Colloquially, the phrase "genetic makeup" is sometimes used to signify the genome of a particular individual or organism. The study of the global properties of genomes of related organisms is usually referred to as genomics, which distinguishes it from genetics which generally studies the properties of single genes or groups of genes.

Both the number of base pairs and the number of genes vary widely from one species to another, and there is only a rough correlation between the two (an observation is known as the C-value paradox). At present, the highest known number of genes is around 60,000, for the protozoan causing trichomoniasis (see List of sequenced eukaryotic genomes), almost three times as many as in the human genome.

An analogy to the human genome stored on DNA is that of instructions stored in a book:

- The book (genome) would contain 23 chapters (chromosomes);
- Each chapter contains 48 to 250 million letters (A,C,G,T) without spaces;
- Hence, the book contains over 3.2 billion letters total;
- The book fits into a cell nucleus the size of a pinpoint;
- At least one copy of the book (all 23 chapters) is contained in most cells of our body. The only exception in humans is found in mature red blood cells which become enucleated during development and therefore lack a genome.

Sequencing and mapping

In 1976, Walter Fiers at the University of Ghent (Belgium) was the first to establish the complete nucleotide sequence of a viral RNA-genome (Bacteriophage MS2). The next year Fred Sanger completed the first DNA-genome sequence: Phage Φ -X174, of 5386 base pairs.^[8] The first complete genome sequences among all three domains of life were released within a short period during the mid-1990s: The first bacterial genome to be sequenced was that of *Haemophilus influenzae*, completed by a team at The Institute for Genomic Research in 1995. A few months later, the first eukaryotic genome was completed, with sequences of the 16 chromosomes of budding yeast *Saccharomyces cerevisiae* published as the result of a European-led effort begun in the mid-1980s. The first genome sequence for an archaeon, *Methanococcus jannaschii*, was completed in 1996, again by The Institute for Genomic Research.

The development of new technologies has made it dramatically easier and cheaper to do sequencing, and the number of complete genome sequences is growing rapidly. The US National Institutes of Health maintains one of several comprehensive databases of genomic information.^[9] Among the thousands of completed genome sequencing projects include those for rice, a mouse, the plant *Arabidopsis thaliana*, the puffer fish, and the bacteria *E. coli*. In December 2013, scientists first sequenced the entire *genome* of a Neanderthal, an extinct species of humans. The genome was extracted from the toe bone of a 130,000-year-old Neanderthal found in a Siberian cave.^{[10][11]}

New sequencing technologies, such as massive parallel sequencing have also opened up the prospect of personal genome sequencing as a diagnostic tool, as pioneered by Manteia Predictive Medicine. A major step toward that goal was the completion in 2007 of the full genome of James D. Watson, one of the co-discoverers of the structure of DNA.^[12]

Whereas a genome sequence lists the order of every DNA base in a genome, a genome map identifies the landmarks. A genome map is less detailed than a genome sequence and aids in navigating around the genome. The Human Genome Project was organized to map and to sequence the human genome. A fundamental step in the project was the release of a detailed genomic map by Jean Weissenbach and his team at the Genoscope in Paris.^{[13][14]}

Reference genome sequences and maps continue to be updated, removing errors and clarifying regions of high allelic complexity.^[15] The decreasing cost of genomic mapping has permitted genealogical sites to offer it as a service,^[16] to the extent that one may submit one's genome to crowd sourced scientific endeavours such as DNA.land at the New York Genome Center, an example both of the economies of scale and of citizen science.^[17]

Genome compositions

Genome composition is used to describe the make up of contents of a haploid genome, which should include **genome size**, proportions of **non-repetitive DNA** and **repetitive DNA** in details. By comparing the genome compositions between genomes, scientists can better understand the evolutionary history of a given genome.

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CATGACGTCGGGACAACCCAGAATTGTCTTGAGCGATGGTAAGTCTAACCTCACTGCCGGGGAGGCTCATAC
CTGGGGCTTACTGATGTCATACCGCTTGCACGGGGATAGAATGACGGTGCCCGGTGTCTGCTTGGCTCGAAGCA
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Part of DNA sequence - prototypification of complete genome of virus

When talking about genome composition, one should distinguish between prokaryotes and eukaryotes as there are significant differences with contents structure. In prokaryotes, most of the genome (85–90%) is non-repetitive DNA, which means coding DNA mainly forms it, while non-coding regions only take a small part.^[18] On the contrary, eukaryotes have the feature of exon-intron organization of protein coding genes; the variation of repetitive DNA content in eukaryotes is also extremely high. In mammals and plants, the major part of the genome is composed of repetitive DNA.^[19]

Most biological entities that are more complex than a virus sometimes or always carry additional genetic material besides that which resides in their chromosomes. In some contexts, such as sequencing the genome of a pathogenic microbe, "genome" is meant to include information stored on this auxiliary material, which is carried in plasmids. In such circumstances then, "genome" describes all of the genes and information on non-coding DNA that have the potential to be present.

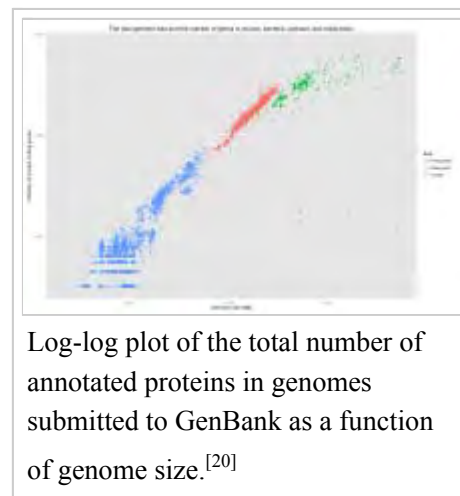
In eukaryotes such as plants, protozoa and animals, however, "genome" carries the typical connotation of only information on chromosomal DNA. So although these organisms contain chloroplasts or mitochondria that have their own DNA, the genetic information contained in DNA within these organelles is not considered part of the genome. In fact, mitochondria are sometimes said to have their own genome often referred to as the "mitochondrial genome". The DNA found within the chloroplast may be referred to as the "plastome".

Genome size

Genome size is the total number of DNA base pairs in one copy of a haploid genome. In humans, the nuclear genome comprises approximately 3.2 billion nucleotides of DNA, divided into 24 linear molecules, the shortest 50 000 000 nucleotides in length and the longest 260 000 000 nucleotides, each contained in a different chromosome.^[21] The genome size is positively correlated with the morphological complexity among prokaryotes and lower eukaryotes; however, after mollusks and all the other higher eukaryotes above, this correlation is no longer effective.^{[19][22]} This phenomenon also indicates the mighty influence coming from repetitive DNA act on the genomes.

Since genomes are very complex, one research strategy is to reduce the number of genes in a genome to the bare minimum and still have the organism in question survive. There is experimental work being done on minimal genomes for single cell organisms as well as minimal genomes for multi-cellular organisms (see Developmental biology). The work is both *in vivo* and *in silico*.^{[23][24]}

Here is a table of some significant or representative genomes. See #See also for lists of sequenced genomes.



Organism type	Organism	Genome size (base pairs)		Approx. no. of genes	Note
Virus	Porcine circovirus type 1	1,759	1.8kb		Smallest viruses replicating autonomously in eukaryotic cells. ^[25]
Virus	Bacteriophage MS2	3,569	3.5kb		First sequenced RNA-genome ^[26]
Virus	SV40	5,224	5.2kb		[27]
Virus	Phage Φ -X174	5,386	5.4kb		First sequenced DNA-genome ^[28]
Virus	HIV	9,749	9.7kb		[29]
Virus	Phage λ	48,502	48kb		Often used as a vector for the cloning of recombinant DNA. [30] [31] [32]
Virus	Megavirus	1,259,197	1.3Mb		Until 2013 the largest known viral genome. [33]
Virus	<i>Pandoravirus salinus</i>	2,470,000	2.47Mb		Largest known viral genome. ^[34]
Bacterium	<i>Nasuia deltocephalinicola</i> (strain NAS-ALF)	112,091	112kb		Smallest non-viral genome. ^[35]
Bacterium	<i>Carsonella ruddii</i>	159,662	160kb		
Bacterium	<i>Buchnera aphidicola</i>	600,000	600kb		[36]
Bacterium	<i>Wigglesworthia glossinidia</i>	700,000	700Kb		
Bacterium	<i>Haemophilus influenzae</i>	1,830,000	1.8Mb		First genome of a living organism sequenced, July 1995 [37]
Bacterium	<i>Escherichia coli</i>	4,600,000	4.6Mb	4288	[38]
Bacterium	<i>Solibacter usitatus</i> (strain Ellin 6076)	9,970,000	10Mb		[39]
Bacterium – cyanobacterium	<i>Prochlorococcus</i> spp. (1.7 Mb)	1,700,000	1.7Mb	1884	

Organism type	Organism	Genome size (base pairs)		Approx. no. of genes	Note
					Smallest known cyanobacterium genome ^{[40][41]}
Bacterium – cyanobacterium	<i>Nostoc punctiforme</i>	9,000,000	9Mb	7432	7432 "open reading frames" ^[42]
Amoeboid	<i>Polychaos dubium</i> ("Amoeba" dubia)	670,000,000,000	670Gb		Largest known genome. ^[43] (Disputed) ^[44]
Plant	<i>Genlisea tuberosa</i>	61,000,000	61Mb		Smallest recorded flowering plant genome, 2014. ^[45]
Plant	<i>Arabidopsis thaliana</i>	157,000,000	157Mb	25498	First plant genome sequenced, December 2000. ^[46]
Plant	<i>Populus trichocarpa</i>	480,000,000	480Mb	73013	First tree genome sequenced, September 2006 ^[47]
Plant	<i>Fritillaria assyrica</i>	130,000,000,000	130Gb		
Plant	<i>Paris japonica</i> (Japanese-native, pale-petal)	150,000,000,000	150Gb		Largest plant genome known ^[48]
Plant – moss	<i>Physcomitrella patens</i>	480,000,000	480Mb		First genome of a bryophyte sequenced, January 2008. ^[49]
Fungus – yeast	<i>Saccharomyces cerevisiae</i>	12,100,000	12.1Mb	6294	First eukaryotic genome sequenced, 1996 ^[50]
Fungus	<i>Aspergillus nidulans</i>	30,000,000	30Mb	9541	[51]
Nematode	<i>Pratylenchus coffeae</i>	20,000,000	20Mb		[52] Smallest animal genome known ^[53]
Nematode	<i>Caenorhabditis elegans</i>	100,300,000	100Mb	19000	First multicellular animal genome sequenced, December 1998 ^[54]
Insect	<i>Drosophila melanogaster</i> (fruit fly)	175,000,000	175Mb	13600	Size variation based on strain (175-

Organism type	Organism	Genome size (base pairs)		Approx. no. of genes	Note
					180Mb; standard <i>y w</i> strain is 175Mb) ^[55]
Insect	<i>Apis mellifera</i> (honey bee)	236,000,000	236Mb	10157	[56]
Insect	<i>Bombyx mori</i> (silk moth)	432,000,000	432Mb	14623	14,623 predicted genes ^[57]
Insect	<i>Solenopsis invicta</i> (fire ant)	480,000,000	480Mb	16569	[58]
Mammal	<i>Mus musculus</i>	2,700,000,000	2.7Gb	20210	[59]
Mammal	<i>Homo sapiens</i>	3,289,000,000	3.3Gb	20000	<i>Homo sapiens</i> estimated genome size 3.2 billion bp ^[60] Initial sequencing and analysis of the human genome ^[61]
Mammal	<i>Bonobo</i>	3,286,640,000	3.3Gb	20000	<i>Pan paniscus</i> estimated genome size 3.29 billion bp ^[62]
Fish	<i>Tetraodon nigroviridis</i> (type of puffer fish)	385,000,000	390Mb		Smallest vertebrate genome known estimated to be 340 Mb ^{[63][64]} – 385 Mb. ^[65]
Fish	<i>Protopterus aethiopicus</i> (marbled lungfish)	130,000,000,000	130Gb		Largest vertebrate genome known

Proportion of non-repetitive DNA

The **proportion of non-repetitive DNA** is calculated by using the length of non-repetitive DNA divided by genome size. Protein-coding genes and RNA-coding genes are generally non-repetitive DNA.^[66] A bigger genome does not mean more genes, and the proportion of non-repetitive DNA decreases along with increasing genome size in higher eukaryotes.^[19]

It had been found that the proportion of non-repetitive DNA can vary a lot between species. Some *E. coli* as prokaryotes only have non-repetitive DNA, lower eukaryotes such as *C. elegans* and fruit fly, still possess more non-repetitive DNA than repetitive DNA.^{[19][67]} Higher eukaryotes tend to have more repetitive DNA than non-repetitive ones. In some plants and amphibians, the proportion of non-repetitive DNA is no more than 20%, becoming a minority component.^[19]

Proportion of repetitive DNA

The **proportion of repetitive DNA** is calculated by using length of repetitive DNA divide by genome size. There are two categories of repetitive DNA in genome: tandem repeats and interspersed repeats.^[68]

Tandem repeats

Tandem repeats are usually caused by slippage during replication, unequal crossing-over and gene conversion,^[69] satellite DNA and microsatellites are forms of tandem repeats in the genome.^[70] Although tandem repeats count for a significant proportion in genome, the largest proportion in mammalian is the other type, interspersed repeats.

Interspersed repeats

Interspersed repeats mainly come from transposable elements (TEs), but they also include some protein coding gene families and pseudogenes. Transposable elements are able to integrate into the genome at another site within the cell.^{[18][71]} It is believed that TEs are an important driving force on genome evolution of higher eukaryotes.^[72] TEs can be classified into two categories, Class 1 (retrotransposons) and Class 2 (DNA transposons).^[71]

Retrotransposons

Retrotransposons can be transcribed into RNA, which are then duplicated at another site into the genome.^[73] Retrotransposons can be divided into Long terminal repeats (LTRs) and Non-Long Terminal Repeats (Non-LTR).^[72]

Long terminal repeats (LTRs)

similar to retroviruses, which have both gag and pol genes to make cDNA from RNA and proteins to insert into genome, but LTRs can only act within the cell as they lack the env gene in retroviruses.^[71] It has been reported that LTRs consist of the largest fraction in most plant genome and might account for the huge variation in genome size.^[74]

Non-long terminal repeats (Non-LTRs)

can be divided into long interspersed elements (LINEs), short interspersed elements (SINEs) and Penelope-like elements. In *Dictyostelium discoideum*, there is another DIRS-like elements belong to Non-LTRs. Non-LTRs are widely spread in eukaryotic genomes.^[75]

Long interspersed elements (LINEs)

are able to encode two Open Reading Frames (ORFs) to generate transcriptase and endonuclease, which are essential in retrotransposition. The human genome has around 500,000 LINES, taking around 17% of the genome.^[76]

Short interspersed elements (SINEs)

are usually less than 500 base pairs and need to co-opt with the LINES machinery to function as nonautonomous retrotransposons.^[77] The Alu element is the most common SINEs found in primates, it has a length of about 350 base pairs and takes about 11% of the human genome with around 1,500,000 copies.^[72]

DNA transposons

DNA transposons generally move by "cut and paste" in the genome, but duplication has also been observed. Class 2 TEs do not use RNA as intermediate and are popular in bacteria, in metazoan it has also been found.^[72]

Genome evolution

Genomes are more than the sum of an organism's genes and have traits that may be measured and studied without reference to the details of any particular genes and their products. Researchers compare traits such as karyotype (chromosome number), genome size, gene order, codon usage bias, and GC-content to determine what mechanisms could have produced the great variety of genomes that exist today (for recent overviews, see Brown 2002; Saccone and Pesole 2003; Benfey and Protopapas 2004; Gibson and Muse 2004; Reese 2004; Gregory 2005).

Duplications play a major role in shaping the genome. Duplication may range from extension of short tandem repeats, to duplication of a cluster of genes, and all the way to duplication of entire chromosomes or even entire genomes. Such duplications are probably fundamental to the creation of genetic novelty.

Horizontal gene transfer is invoked to explain how there is often an extreme similarity between small portions of the genomes of two organisms that are otherwise very distantly related. Horizontal gene transfer seems to be common among many microbes. Also, eukaryotic cells seem to have experienced a transfer of some genetic material from their chloroplast and mitochondrial genomes to their nuclear chromosomes.

See also

- Bacterial genome size
- Cryoconservation of animal genetic resources
- Genome Browser
- Genome Compiler
- Genome topology
- Genome-wide association study
- List of sequenced animal genomes
- List of sequenced archaeal genomes



- List of sequenced bacterial genomes
- List of sequenced eukaryotic genomes
- List of sequenced fungi genomes
- List of sequenced plastomes
- List of sequenced protist genomes
- Metagenomics
- Microbiome
- Molecular epidemiology
- Molecular pathological epidemiology
- Molecular pathology
- Nucleic acid sequence
- Pan-genome
- Precision medicine
- Sequenceome
- Whole genome sequencing

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- UCSC Genome Browser (<http://genome.ucsc.edu>) – view the genome and annotations for more than 80 organisms.
- genomecenter.howard.edu (<http://www.genomecenter.howard.edu/>)
- Build a DNA Molecule (<http://learn.genetics.utah.edu/content/begin/dna/builddna/>)
- Some comparative genome sizes (http://www.genomenetwork.org/articles/02_01/Sizing_genomes.shtml)
- DNA Interactive: The History of DNA Science (<http://www.dnai.org/>)
- DNA From The Beginning (<http://www.dnaftb.org/>)
- All About The Human Genome Project (<http://www.genome.gov/10001772>)—from Genome.gov
- Animal genome size database (<http://www.genomesize.com/>)
- Plant genome size database (<http://www.rbgekew.org.uk/cval/homepage.html>)
- GOLD:Genomes OnLine Database (<http://www.genomesonline.org/>)
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- NCBI Entrez Genome Project database (<http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?db=genomeprj>)
- NCBI Genome Primer (http://www.ncbi.nlm.nih.gov/About/primer/genetics_genome.html)
- GeneCards (<http://www.genecards.org/>)—an integrated database of human genes
- Visualization of nucleotide sequence (<http://studia.scienceontheweb.net/visualization.php>) - prototypification of complete genome of virus, sequence of 5418 nucleotides
- BBC News – Final genome 'chapter' published (<http://news.bbc.co.uk/1/hi/sci/tech/4994088.stm>)
- IMG (<http://img.jgi.doe.gov/>) (The Integrated Microbial Genomes system)—for genome analysis by the DOE-JGI
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