

Genotype

The term **genotype** means either information about the genetic constitution **of a cell**, or **an organism** and/or **an individual**. The genotype of a specific individual then represents all of its genetic characteristics. Genotypes of individuals of the same species differ in terms of genomic sequences in genic and extragenic regions. One example of high variability of genotypes between individuals of the human population is, for example, the HLA locus.

According to current knowledge, the genotype represents both a set of genes and the number of copies of genes (see e.g. rRNA genes) or smaller or larger regions of the genome (CNV – copy number variation), polymorphisms in the nucleotide **sequence** (such as SNP – single nucleotide polymorphism; indel – **insertion** or **deletion**), or variability in the length of repetitive sequences. However, it also includes other characteristics of the genome, such as genomic imprinting (see Epigenetics).

The genetic make-up of an individual (with the exception of gametes) is diploid. A genotype in the narrower sense is therefore a pair of alleles of the same gene, where one allele from the pair is of maternal origin and the other is of paternal origin. E.g. the human gene encoding phenylalanine hydroxylase exists in the human population in two allelic forms: dominant *A* and recessive, *and* it follows that there are three possible genotypes in the human population – *AA* (dominant homozygotes), *Aa* (heterozygotes) and *aa* (recessive homozygotes).

Determining an individual's genotype, genotyping, is made possible by various biological procedures (genotyping assays), such as PCR polymerase chain reaction, RFLP restriction fragment length polymorphism analysis, sequencing, and others.

Links

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