

# Structure and function of a gene

A gene is the basic unit that determines the characteristics of an individual. Genes are a means of transmitting hereditary information from a parent cell to a daughter cell. Genetics has been dealing with this issue since the beginning of the 20th century. In the 1940s, it was discovered that the main purpose of genetic information is to create proteins (enzymes, structural components, regulators of gene expression,...) In recent years, the definition of a gene has been modified: "A gene is a localized region of a genomic sequence, corresponding to a unit of heredity, which is associated with regulatory and transcriptional regions".

The main carrier of genetic information is deoxyribonucleic acid (DNA). The main mechanism of DNA copying was explained in 1953 by **James Watson** and **Francis Crick**. Genes are located on chromosomes, which was proved in 1944. The region of the chromosome where a given gene is located is called a locus. A chromosome is made up of one very long DNA helix, on which hundreds to thousands of genes are coded.

## Gene structure

The information in DNA is determined by the sequence of individual nucleotides. Biological information is written using the bases - **adenine** (A), **cytosine** (C), **guanine** (G) and **thymine** (T). Genes contain instructions for the creation of proteins. The linear sequences of nucleotides must therefore be related with coding of the given amino acid. It took 10 years to clarify the relationship between the four-letter base system and the twenty-one-letter amino acid alphabet.

Most genes are made up of relatively short coding sections of DNA (exons). Genes are dominated by non-coding sequences (introns), which are later excised. The complete set of all genetic information of an organism is called a genome.

The amount of genetic information in a cell is enormous. The human genome contains around  $3 \times 10^9$  nucleotides (corresponding to approximately 1 meter in length). The complete nucleotide sequence of the human genome would then take up around 900,000 pages. All this information must be copied during cell division, hence errors may occur. For this reason the cell contains repair mechanisms.

Most of the genes in the cell are located **in the nucleus on chromosomes** – nuclear genes (they are critically important for the life of an individual and its species). Genes stored in chromosomes of cytoplasmic structures are called **plasmagenes**.

## Introns and exons

- in higher organisms, including humans, the **coding information of a gene** is organized into a series of DNA segments called **exons**
- exons are separated by sections of non-coding sequences – **introns**
- the number and length of exons and introns varies widely, but **the length of introns** is usually much greater than the length of exons

## Regulatory Areas

- **regulatory regions** that control the initiation or cessation of a certain process, for example **the expression of genetic information**, are part of the gene
- here, the so-called **promotor** is of fundamental importance, a section of DNA that is placed towards the 5' end of the DNA strand from the place of **the start of transcription** (the term upstream is used of vice versa for downstream sequences – **downstream** towards the 3' end)
- the promotor contains specific so-called **signal sequences**, which are recognized by transcription factors (proteins) and through them by **RNA-polymerase**
- depending on their binding, **transcription of the gene is subsequently initiated (or stopped)**
- these signal sequences are highly **conserved**, meaning they are the same or similar across animal species
- it is related to their **important functions**, as mutations of signal sequences have serious consequences for the cell and are therefore classified as **forbidden mutations** from an evolutionary point of view (mutational evolutionary mechanisms)

**The size of genes** varies, ranging from less than **100bp** (bp-base pairs) to several **million bp**.

- most genes are **unevenly** distributed on chromosomes, however some exist in **groups (clusters)**
- these genes are more or less similar and form so-called **gene families**
- gene families arose in the course of evolution by the mechanism of **repeated duplications** of the original gene and subsequent differentiation due to mutations

## Functions of genes

Gene expression is initiated by the process of transcription, when DNA is transcribed into RNA (it contains uracil instead of thymine). RNA is single-stranded and less stable than DNA. Individual codons (triples of nucleotides carry information about the amino acid arrangement of the emerging proteins.

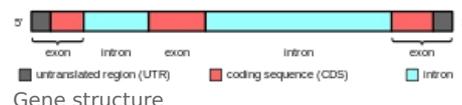
Transcription and translation is influenced by regulatory regions of the genome. The area where the gene is transcribed is decided by the promotor. For one gene, we can encounter a larger number of promoters. Genes contain both coding and non-coding regions – exons and introns.

## Expression of genetic information

The process by which this information becomes usable by the cell is called **gene expression**.

**Crick** described this process as **the central dogma of the transfer of genetic information** in the direction of DNA - RNA - protein or DNA -DNA.

- this simple diagram represents a very **complex proces**, which is the result of the regulatory activities of a whole range of **proteins**
- it still needs to be expanded with the possibility of **reverse transcription** – the transfer of information from RNA to DNA, which was first observed in **retroviruses**
- however, it was discovered that even **eukaryotic cells** contain DNA sequences that encode **reverse transcriptase enzymes**
- **reverse transcription** of mature mRNA produces so-called cDNA (complementary DNA)



## Links

### Related Articles

- Gene
- Genetics
- DNA (nucleic acid)
- Chromosomes
- Genome
- Protein-forming and non-protein-forming DNA sequences
- Translation
- Transcription

### References

1. Pearson H (2006). "Genetics: what is a gene?". Nature 441 (7092): 398–401.

### Resources

- Functional structure of a gene ([https://en.wikipedia.org/wiki/Gene#Functional\\_structure\\_of\\_a\\_gene](https://en.wikipedia.org/wiki/Gene#Functional_structure_of_a_gene))

### Used literature

- ALBERTS, B, D BRAY a A JOHNSON. Základy buněčné biologie. 2. vydání. Espero Publishing, 2005. 740 s. ISBN 80-902906-2-0.