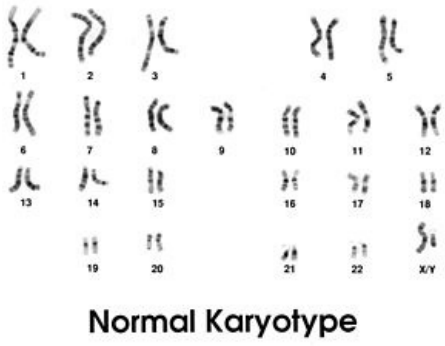
The human genome and its variability

The human genome is **the collection of alll DNA in a human cell**. The genetic information occurs inside the nucleus or outside the nucleus (mitochondria, repetitive sequences).

Nuclear genome



The human nuclear genome (karyotype)

Approximately **3×10^9 base pairs (bp)** are stored in the nucleus in two copies (diploid cells). Each monochromatid chromosome contains one DNA molecule, which is made up of approximately **55 - 250 Mb**[1].

The largest human chromosome is chromosome 1, which is 250 Mb in size, equivalent to a molecule length of 15 cm when unpacked. If we were to transcribe all the genetic information stored in the nucleus into books, we would end up with a library containing a thousand books the size of the Bible.

Genes

Genes Genes are stretches of DNA that encode either a protein or a type of RNA. They are, of course, the most important part of the human genome, as they contain essential information for the life of an organism. Current estimates of the total number of human genes are about **20 000 - 25 000**.[1] They are **unevenly distributed** in chromosomes. They are unevenly distributed in chromosomes. Some chromosomes have a much higher gene density than others (e.g., chromosomes 19 and 22 have a higher gene density, while chromosomes Y and 18 have a relatively low gene density). Individual genes are separated from each other by non-coding sequences. **The coding part of the genome makes up approximately 3%**[1] of the total DNA. The exact function of the non-coding sequences is unknown.

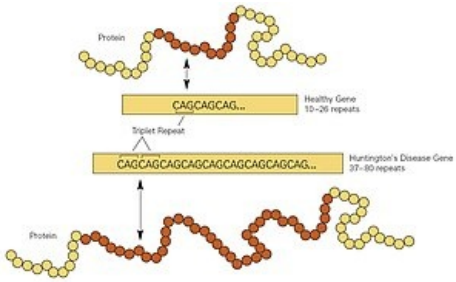
Some genes are found in **multiple copies** - called gene families. These gene families are made up of genes with similar sequence, structure and function. Their **evolutionary function**. [1] (https://en.wikipedia.org/wiki/Susumu_Ohno) (e.g. there are 10-100 copies of genes for tRNA synthesis, and up to 50 copies for histones).

Pseudogenes

Pseudogenes (sometimes also gene fragments) are remnants of genes that have **lost their significance** during evolution. Jelikož nekódují žádný znak, nahromadily se v nich během vývoje mnohé mutace (neboť nepůsobil žádný selekční tlak proti mutacím). Since they do not encode any trait, they have accumulated many mutations during evolution (since no selection pressure against mutations has acted). In some cases, their similarity to the original gene can **complicate genetic tests**.

Extragenic DNA

Extragenic DNA includes **non-coding sequences** that are not part of structural genes, their regulatory regions or distant regulatory elements. This portion of genes makes up approximately**70-80 %**[1] of the human genome.



Repetitive sequences (Huntington's chorea)

Repetetive sequences

DNA sequences of varying length that occur in many copies. They make up approximately **50 %**[1] **of non-coding DNA**.

🔎 *For more information see Repetitive sequences in the human genome.*

Mitochondrial DNA

The mitochondrial genome (mtDNA) is a structure that is about 93 %[1] made up of coding sequences. This is mainly due to the absence of introns. The genes are arranged closely together. Some coding sequences overlap. Mitochondrial DNA is inherited maternally.

🔎 *For more information see Genetic makeup of mitochondria.*

Human genome variability

The general name for different sequences in certain parts of the genome is variation, but nowadays the term **polymorphism** is more commonly used. When it comes to variants within human genes, mutations in exons, as well as introns or promoter sequences can cause disease.

🔎 *For more information see Nucleic acid polymorphism.*

Links

Related articles

- Gene
- Nucleic acid polymorphisms
- Repetitive sequences in the human genome
- Mitochondrial DNA

References used

- KOHOUTOVÁ, Milada. *Medical biology and genetics (II. part).* 1. edition. Prague : Karolinum Publishers, 2013. 202 pp. ISBN 978-80-246-1873-9.

References

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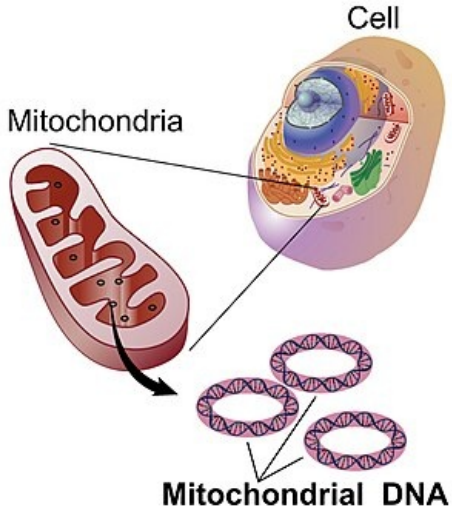

Mitochondrial DNA