Genotype and its variability, mutation and recombination

Genotype

Genotype is a specific *combination of hereditary dispositions*, a *set of all alleles* (forms of genes) of an individual. It could also be defined as the *genetic constitution of an individual*, or more specifically: *a specific set of alleles* at a specific locus or multiple loci.

- represents the genetic constitution of an organism represented by a set of alleles specifically arranged in the genome (therefore, they are all DNA or RNA molecules (in the case of RNA viruses) of a living system that are characterized by replication and are passed on to offspring)
- determines the extent and degree of phenotypic (set of all characters) possibilities of an individual

Mutation

The DNA sequence is subject to changes that are **caused** by the action of chemical, physical and biological agents or arise as **rare errors** during replication (endogenous mutations) - resulting in **allelic variations**.

- **random hereditary changes** in the genotype (genetic information) are called mutations
- mutations resulting from errors in DNA replication are called **spontaneous mutations** and occur without intervention from the external environment
- **DNA polymerase** is not only very accurate, but also has a self-correcting function – the probability of an error is in the order of about 10^{-7}
- the frequency of mutations is therefore **very low**, moreover, cells are to some extent capable of eliminating these errors thanks to repair enzymes
- most mutations are therefore so-called **induced**, i.e. induced by *external mutagenic factors* (mutagens) – these can be, for example, radiation (UV, X-rays), chemical substances (arenes, heavy metals, peroxides...) etc.
- mutations can occur to varying extents:

Gene point mutation

They take place at the level of the DNA molecule, affecting **1 gene**. The result is a damaged nucleotide sequence, due to which *triplets (codons) are changed* and an error occurs in proteosynthesis (completely different amino acids are synthesized).

If the **gene regulating the multiplication and differentiation** of the cell is damaged, it can lead to uncontrollable *growth* (tumorous diseases).

It can happen:

- **substitution** - the exchange of one nucleotide for a nucleotide carrying another base, is among the most frequently occurring mutations
  - **transition** – exchange of purine-purine, pyrimidine-pyrimidine
  - **transversion** – exchange of purine – pyrimidine and vice versa
- **deletion** - loss of one or more nucleotides in a DNA sequence
- **insertion** – one or more nucleotides are inserted into the DNA sequence

Distribution of gene mutations in terms of **effect on the gene product**: both **coding and non-coding** sequences of the human genome can be subject to mutations, mainly mutations related to **coding DNA** have serious consequences

- **synonymous (silent)** – does not change the sequence of the gene product
- **nonsynonymous** - changes the sequence of a gene product (protein or RNA) with all possible consequences

Chromosomal mutation (chromosomal aberration)

There is a **change in the number or structure** of chromosomes - for example, during *crossing over*: chromosome fragments do not connect properly; but the entire block can also be lost.

These mutations *disrupt the course of meiosis* and cause gametes to become *non-functional*.

Types:

1. **deletion** (loss of part of a chromosome)
2. **inversion** (turning over part of a chromosome)
3. **duplication** (doubling of part of a chromosome)
4. **translocation** (attachment of part of the chromosome to the wrong chromosome)
5. **fragmentation** (breakdown of the chromosome into fragments)

Genomic mutations

There is a change in the **genome itself**. Mostly it is either a multiplication of the entire chromosome set (*euploidy = polyploidy* = an individual has 3n or more...), or a change in the number of individual chromosomes from the set (*aneuploidy*).

Inversion of part of a chromosome

Mutations can be distinguished according to the type of **affected cells**:

1. **somatic** – affect the offspring of the mutated cell, but are not transmitted between individuals
2. **gametic** - affect cells of the germline, can be transmitted from parents to offspring

Mutations can be distinguished according to the **mechanism of origin**:

1. **spontaneous**
2. **induced**

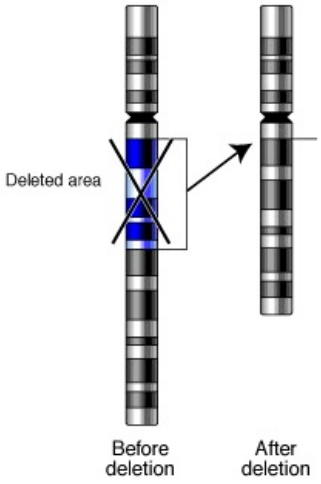
Recombination

Recombination is the name for various changes in DNA, consisting in splitting it and connecting it to another chain, which gives rise to **new properties**

1. **Meiotic recombination** – i.e. crossing – over
   - the process during which **two homologous chromosomes** paired in prophase I of meiosis **exchange part of their DNA**, the result of a correctly performed crossing-over is the exchange of part of the **alleles between chromosomes** - i.e. disruption of gene linkage
   - the consequence is a considerable increase in the **variability of the offspring**

Crossing-over is one of the main sources of **genetic variability**, in addition to mutations and the random separation of chromosomes into gametes, it does not create new alleles, but allows the creation of **new combinations** of already existing alleles of genes located on the **same chromosome** (see gene linkage)

- we distinguish between **simple crossing-over** (one crossing over occurs, chromatids swap ends) and **multiple crossing-over** (several crossed over, sections "inside" chromatids are also swapped)
- **multiple crossing-overs** disturb the calculations determining the strength of linkage and the distance of genes on the chromosome - a three-point test is used to filter them out



deletion of part of a chromosome

1. **Mitotic recombination** – very rare
   - **recombinant DNA technology** - a procedure in genetic engineering in which simple genes are isolated from cells and then introduced back into cells of the same or different species of organism

- these are **biotechnological procedures** that make it possible to create new combinations of DNA molecules that do not occur together in a natural organism
- recombinant DNA technology is used, for example, in gene therapy or **genetic modification** (GMO - e.g. modified corn or soy)
- The prerequisite for evolution is **variability**. Genetic variability is increased by mutation, recombination and gene flow. Recombination is the **rearrangement of DNA material** between already existing alleles (forms of genes) to create new alleles. This process creates new individuals with combinations of alleles different from those of their parents. This happens due to *free combinability* or *crossing-over*.

Links

Related articles

- Genotype
- Mutation
- Crossing-over
- Gametogenesis

Sources

- ŠTEFÁNEK, Jiří. *Medicína, nemoci, studium na 1. LF UK* [online]. [cit. 11.02.2010]. <http://www.stefajir.cz>.