

## Lecture 2. Structural genomics. Human genome.

### Learning outcomes:

1. Give the definition to the following terms: "gene", "genotype", "genome", "genetics" and "genomics".
2. Characterize the differences between structural and functional genomics, give the definitions.
3. Describe the history of the Human Genome Project. What is the meaning of this project?
4. Analyze the applications of the results of this project in medicine and pharmacy, their future perspectives.
5. Briefly describe the structure of human genome.

The **gene** is the **hereditary factor** carrying the information about specific trait or function of the organism and it is the structural and functional unit of heredity. The **genotype** is only **the set of genes** of living organism. **Genetics** is the science about the regularities of **heredity** and **changeability**. **Genomics** is an interdisciplinary field of biology focusing on the structure, function, evolution, mapping, and editing of **genomes**. A **genome** is an organism's **complete set of DNA**, including all of its **genes** and **intergenic regions** called "spacers". In contrast to genetics, which refers to the study of *individual genes* and their roles in inheritance, genomics aims at the collective characterization and quantification of all of an organism's genes, their interrelations and influence on the organism. **Structural genomics** seeks to describe the **3-dimensional structure of every protein** encoded by a given genome. **Functional genomics** is a field of molecular biology that attempts to make use of the vast wealth of data produced by genomic projects (such as genome sequencing projects) to describe **gene (and protein) functions and interactions**. Functional genomics focuses on the dynamic aspects such as **gene transcription, translation and protein-protein interactions**, as opposed to the static aspects of the genomic information such as DNA sequence or structures.

The Human Genome Project (HGP) started in 1990 and finished in 2003. As the result of this project it was found that the human genome contains 3.2 billion of base pairs and approximately 25-30 thousands of protein-coding genes on 46 chromosomes of nuclear DNA. The genome of human mitochondrial DNA contains only 16.5 thousands of base pairs and only several specific genes.

### The questions for self - control:

1. What are the gene, the genotype, the genome, the genetics and the genomics?
2. What are the structural and functional genomics, what is the difference between them?
3. The history and medico-biological meaning of Human Genome Project.
4. What is the human genome, what is its structure and which components it contains?

### Recommended readings:

1. "WHO definitions of genetics and genomics". World Health Organization.
2. Marsden RL, Lewis TA, Orengo CA (March 2007). "Towards a comprehensive structural coverage of completed genomes: a structural genomics viewpoint". BMC Bioinformatics. 8: 86. doi:10.1186/1471-2105-8-86. PMC 1829165. PMID 17349043.
3. Brenner SE, Levitt M (January 2000). "Expectations from structural genomics". Protein Science. 9 (1): 197–200. doi:10.1110/ps.9.1.197. PMC 2144435. PMID 10739263.
4. McElheny VK (2010). Drawing the Map of Life: Inside the Human Genome Project. Basic Books. ISBN 978-0-465-03260-0. 361 pages. Examines the intellectual origins, history, and motivations of the project to map the human genome; draws on interviews with key figures.