# Level: PhD

Course title: Human molecular biology and genetics (DSB620)

### Status: elective

**ECTS**: 15

## **Requirements**:

## Learning objectives

The main objective of the course is to enable the PhD students to acquire knowledge and skills about the organization of the human genome, molecular biology methods, molecular basis of the heritable diseases and application of molecular biology methods in human diseases diagnostics. Students will acquire the comprehensive knowledge about the new and modern methods and techniques applied in research of structure and variations of the human genome, about the expression of genetic information and mutations of human genome, with the main aim to apply the acquired knowledge in research.

# Learning outcomes

PhD students will acquire comprehensive knowledge about the methodology and application of molecular biology techniques in research of genetic basis of human diseases and about the bioinformatics and technological advances in human genome research, including the human genome variations at the individual and population level.

# Syllabus

Theoretical instruction

- 1. Human genome structure
- 2. Functional organization of the human genome
- 3. Chromosomal basis of inheritance
- 4. Cell cycle, DNA replication, transcription, translation
- 5. Gene expression regulation
- 6. Mutation, DNA reparation and recombination
- 7. Molecular biomarkers
- 8. Methods of molecular biology and human genetics
- 9. Principles of cytogenetics in human disease diagnostics
- 10. Genetics of heritable and multifactorial diseases
- 11. Genetic variations in medical genetics and population genetics
- 12. Gene mapping and association studies
- 13. Prenatal diagnostics
- 14. Malignant diseases genetics and genomics
- 15. Pharmacogenomics
- 16. Ethical aspects of genetics and molecular biology

### Practical instruction

- 1. Methodology of molecular biology
- 2. Methods of human genetics
- 3. Numerical and structural chromosomal aberrations
- 4. Polymerase chain reaction
- 5. Isolation and identification of genes
- 6. Bioinformatics in molecular biology and human genetics
- 7. Methods for detection of polymorphisms and mutations
- 8. Vectors and cloning
- 9. Basics of personalized and translational medicine

Weekly teaching load: 10 (150)			Other:
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