

Course title: MOLECULAR DIAGNOSTICS

ECTS credit allocation (and other scores): 6

Semester: autumn

Level of study: ISCED-7 - second-cycle programmes (EQF-7)

Branch of science: Natural sciences

Language: English

Number of hours per semester: 75 h.

Course coordinator/ Department and e-mail: Nina Smolińska; Department of Animal Anatomy and Physiology;
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Type of classes: classes and lectures

Substantive content

CLASSES: Practical application of selected methods in the diagnosis of various genetic markers. Examination of the scene. Detection, preservation and collection of genetic material at the crime scene. DNA isolation from evidence and comparator. Isolation of own gDNA - as a matrix for medical diagnostics. Molecular diagnosis of sex based on amelogenin gene amplicon length. Identification of STR short tandem repeat polymorphisms in the human genome (FGA, VWA, or CD4 alleles). Forensic individualization based on the diagnosis of the length of amplicons of selected genetic markers. Diagnostics of selected genes by the evaluation of restriction fragment length polymorphism (PCR-RFLP). Identification of selected genetic mutations (e.g. detection of deletions in the human CCR5 receptor gene determining resistance to HIV). Detection of cytomegalovirus by nested PCR. Identification of the polymorphism of the alcohol dehydrogenase gene (ADH3) by the PCR-RFLP method. Identification of insertion-deletion polymorphism of the angiotensin converting enzyme (ACE) gene. Correct analysis and evaluation of the obtained results. Seminar – “Application of molecular biology in forensics and molecular diagnostics of a selected genetic disease”.

LECTURES: Biological traces depending on the type of forensic events. DNA profile databases. Genetic expertise (selection of research methodology) depending on the type of trace. Principles of source selection and quality assessment of gDNA matrix used for proper molecular forensic and medical diagnostics. Molecular forensic diagnostics: individual identification based on polymorphic markers (STR, VNTR, SNP), gender identification (amelogenin gene, SRY, ZFX, ZFY) and paternity determination. Legal regulations for the functioning of forensic molecular biology (ENFSI recommendations - European Network of Forensic Science Institutes, The treaty of Prum). Laboratory accreditation - ISO 17025. Tests of competence. Molecular medical diagnosis of genetic diseases (e.g. chromosomal aberrations, Marfan syndrome, cystic fibrosis, phenylketonuria, porphyria, muscular dystrophy, mitochondrial diseases, hypertension, breast cancer). Invasive and non-invasive prenatal diagnosis - advantages and disadvantages. Prevention of genetic diseases. Gene therapy.

LEARNING PURPOSE: Getting to know the basic methods used in individualized forensic diagnostics and molecular diagnostics of the most common genetic diseases.

On completion of the study programme the graduate will gain:

KNOWLEDGE: the student knows and understands the molecular biology methods used in molecular diagnostics forensic and medical, classical and molecular diagnostic methods used in biological research, the basic principles of ergonomics, hygiene and occupational safety with biological material.

SKILLS: the student can use advanced techniques and research tools used in molecular biology, operate research equipment used in biological sciences (for research at the molecular and cellular level), use the extended theoretical knowledge that allows for the description and explanation of processes and phenomena occurring in nature and everyday

life, as well as specialist knowledge in the field of study) in the analysis and extrapolation of biological processes, determining the correctness and formulating conclusions.

SOCIAL COMPETENCIES: the student is ready to improve qualifications in the form of education continuous formal and informal development of one's own professional skills, is ready to cooperate and work in a group, assuming various roles and properly prioritizing tasks and projects, disseminating the principle of strict, based on empirical data, interpreting biological phenomena and processes.

Basic literature: 1. Lela Buckingham Molecular Diagnostics: Techniques and Applications for the Clinical Laboratory Elsevier 2015 2. William C. Cho Molecular Pathology: The Diagnostic Challenge Wiley-Blackwell 2012 3. Michael A. M. DeCotiis, Philip G. W. S. Simons Principles of Molecular Medicine Humana Press 2005

Supplementary literature: 1. Guide Ryan J. Smith, Lisa A. Jones Molecular Diagnostics: A Comprehensive Academic Press 2020

The allocated number of ECTS points consists of:

Contact hours with an academic teacher: 79 h.

Student's independent work: 71 h.