University: *University of Presov*

Faculty/university workplace: *Faculty of Humanities and Natural Sciences*

Code: 2BIO/EROSJPRGC/22	**Course title:** *Practice of Human Genetics*

Type, scope and method of educational activity:
Type of educational activity: *Practical seminar*
Scope of educational activity: *2 per semester*
Method of educational activity: *Attendance*

Number of credits: *4*

Recommended semester:

Spring	Biology

Study grade: Master

Prerequisites:

Conditions for passing the course:
Form of assessment:

Continuous evaluation:
The course will be taught in 4 blocks. Each block will last 6 hours and will include a theoretical lecture, explanation of the methodology and planning of the laboratory protocol and the laboratory exercise itself in the laboratory. At the end of the block exercise there will be a written test verifying the understanding of the methodology and mastery of working with databases and software. The condition for admission to the written examination will be active participation in all exercises and submission of the correct protocol from the laboratory exercise
Success criteria: A: 100.00 – 90.00%, B: 89.99 – 80.00%, C: 79.99% - 70.00%, D: 69.99 % - 60.00 %, E: 59.99 % - 50.00 %. FX: 49.99 % and less %.
Final evaluation:
Continuous assessment

Learning outcomes:
Knowledge gained:
Graduate of the course:
- knows pipetting techniques,
- knows, can explain and describe the principle and methodology of electrophoretic methods as well as the importance of individual steps,
- knows, can explain and describe the principle and methodology of PCR methods as well as the importance of the individual steps,
- knows, can explain and describe the principle and methodology of sequencing as well as the significance of the individual steps.

Skills acquired:
Upon completion of the course, the student will be able to plan and carry out a simple experiment involving the detection of mutations in a specific human gene by Sanger sequencing due to the following acquired skills:
- be able to independently perform basic work in a molecular genetic laboratory,
- can operate simple laboratory equipment,
- is able to navigate human genome databases and search them for information necessary for sequencing,

- can design primers,
- be able to carry out the whole process of sample preparation for sequencing, including preparation of the PCR reaction, gel electrophoresis of the product, enzymatic purification of the product, preparation of the sequencing reaction and washing of the product,
- be able to analyse sequencing results using software and find variants in the resulting sequence.
Competences acquired:
The student:
- can creatively solve problems and implement the application of acquired knowledge in research,
- has developed competences for further self-education
- the acquired knowledge, skills and abilities enable the student to continue his/her studies and to apply them in practice.

Course content:
The course will be taught in 4 blocks, each block will take place in one day and will include a theoretical lecture, explanation of the methodology and planning of the laboratory protocol and the laboratory exercise itself in the laboratory.
Day 1: Principle and methodology of sequencing. PCR reaction pipetting.
Day 2: Working with human genome sequence databases - types of databases, searching for polymorphisms and mutations, downloading reference sequences. Students work on PC.
PCR product control by gel electrophoresis. Enzymatic purification of the product and preparation of the sequencing reaction.
Day 3: Primer design. Students work on PC. Principle of technology and work with genetic analyzer AB 3500. Washing of sequencing reaction product. Starting the genetic analyzer.
Day 4: Analysis of sequencing results. Working with sequencing software. Comparison of the resulting sequence with reference sequences. Students work on PC.
Day 5: Submission of protocol from laboratory exercise. Written examination.

Recommended literature :
Sanger sequencing method. 2014 [online]. In: Lifetechnologies [cit. 2014-03-04]. Dostupné na internete: http://www.lifetechnologies.com/sk/en/home/life-science/sequencing/sanger-sequencing/sanger_sequencing_method.html

Notes:

Course evaluation:
Total number of students evaluated:

A	B	C	D	E	FX

Lecturers:
prof. RNDr. Jarmila Bernasovská, PhD., guarantor
doc. RNDr. Dana Dojčáková, PhD., co-guarantor, lecturer, examiner, instructor, seminary supervisor

Date of last change: *01.09.2022*

Approved by: *prof. RNDr. Jarmila Bernasovská, PhD.*