



Course Regime

Course: Practical Bioinformatic Approaches in Medicine

Study Programme:
Medicine

Year of the Course: 2 3 4 5 6

Semester:
Summer

Course type:
Elective

Number of ECTS credits: 3

Lecturer(s): Petra Hudler

Participating Organisational Units (Departments and Institutes): Institute of
Biochemistry

Parts (Modules) of the Course: /

Date of Issue: 15. 9. 2018

A. General part (*applies to compulsory and elective courses*)

1. Course objectives

The objective of the program is to acquaint the students with the basics of research in molecular biology of the disease, for example cancer, leading to translation into clinical diagnostics. The lectures and seminars will provide a solid foundation of information enabling participants to identify candidate genes and design simple experiments. They will learn to use basic web-based tools and databases. They will be familiarized with the bioinformatic approaches used in modern genetics and they will learn how to interpret the results. Students will actively participate in the course by using *in silico* approaches to identify candidate polymorphisms for a model disease and using approaches to identify functional effects of genetic variants on protein function.

2. Comprehensive outline of the course organisation

Lecture room: Seminar room, Institute of Biochemistry, 2. floor

Study activities: introductory lectures, lecture, interactive lectures (computers with internet connection will be available in limited number); cooperative learning; supervision of students preparing their seminar works; Inquiry-Based Learning - students play an active and participatory role in their own learning process. Lecture summaries, study materials and materials for the preparation of the seminars will be available in the online classroom "Spletna učilnica UL MF".

Time: In March, April or May, meeting dates will be finalized in agreement with all participants.

Students' obligations:

- Attendance at seminars is obligatory. Attendance at lectures: 80 %.
- Replacing missing obligations in the case of justified absence: the student must select additional tasks, prepare additional seminar theme (in addition to obligatory seminar) and present the theme to the students, if possible, and the lecturer.
- Seminar: Students should select a seminar topic - problem task in agreement with the subject teacher during lectures. More detailed instructions on how to prepare a seminar will be presented during the final lecture. Students prepare a solution/solutions to the problem task and prepare a short written report according to the instructions published in the UL MF Web Classroom. They should also present their theme to the fellow students and lecturer. The oral presentation is expected to last 5-10 minutes. All students are expected to participate in discussions.
- Instructions for creating a Seminar written report — must contain a short introduction, objective, presentation of results, discussion and conclusion.

3. Description of on-going assessment of knowledge and skills

There are no on-going assessments of knowledge and skills.

4. Required conditions for the final examination (Course Exam)

The student must prepare a written seminar report (elements of the evaluation: length of the presentation, delivery, knowledge and content, clarity of the results, clarity of the conclusions) and defend the seminar at seminar lectures. The students will also be evaluated based on their active participation in discussions.

The student can fulfil the requirements for taking a course exam if he/she has not fulfilled them in the scope of the organised course activities during the course in the current academic year by preparing additional tasks and present them to the lecturer (and students if possible).

5. Final assessment and examination of knowledge and skills (Course Exam)

Presentation of seminar papers (50 % of the grade).

Written seminar report (50 % of the grade).

Evaluation method:

Grading system 1-10 (grades 1-5 unsatisfactory: knowledge does not reach minimum standards, grades 6-10 pass) in accordance with the Regulations for assessment of knowledge and skills for the uniform master's study programs of medicine and dental medicine, which is available on the web site of Medical Faculty.

6. Other provisions

Maximal number of participants at the course: 24.

Eligibility of enrolment to the course: students must have completed courses of Concepts in Biochemistry and lab course of Medical Biochemistry and Molecular Biology.

Preparation of Seminar:

- Seminar: problem-based-learning approach, the student prepares a short report, orally presents the theme (5-10 minutes), discussion.
- Instructions for creating a Seminar written report: must contain a short introduction, objective, presentation of results, discussion and conclusion.

7. Fundamental study material and Supplement reading

Primary study material:

Lectures and lecture notes.

Supplement reading:

1. Collins, F.S. and H. Varmus, A new initiative on precision medicine. *N Engl J Med*, 2015. 372(9): p. 793-5.
2. Kumar, P., S. Henikoff, and P.C. Ng, Predicting the effects of coding non-synonymous variants on protein function using the SIFT algorithm. *Nat Protoc*, 2009. 4(7): p. 1073-81.
3. Landrum, M.J., et al., ClinVar: public archive of interpretations of clinically relevant variants. *Nucleic Acids Res*, 2016. 44(D1): p. D862-8.
4. McLaren, W., et al., The Ensembl Variant Effect Predictor. *Genome Biol*, 2016. 17(1): p. 122.
5. Ritchie, G.R., et al., Functional annotation of noncoding sequence variants. *Nat Methods*, 2014. 11(3): p. 294-6.

8. Exam topics, clinical presentations and skills

Student must be familiar with selected online tools, databases and programs that enable the identification of critical genes involved in complex diseases.

The student must be able to design research methodology or approaches to identify candidate genes in selected diseases.

Knowledge and independent use of the selected web tools and databases that enable bioinformatic determination of cell pathways and interactions between selected genes, in silico determination of functional effects of genetic variability in coding areas, and in silico functional analysis of polymorphisms in regulatory and promotional regions.
Knowledge of approaches for the selection and validation of bio-markers of the disease.

9. Other information

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B. Elective Courses (*considered as Elective Course Announcement*)

1. **Participating main and guest lecturers:** Petra Hudler
2. **Estimated time period in the semester:** March, April, or May
3. **Maximum number of students for the elective course (if the number of students able to attend the course is limited):** 24
4. Please specify if the elective course is available in English for incoming international students (Erasmus + and others). Please specify any additional conditions in the case that the elective course is available for visiting students.

Yes, the elective course is available in English for incoming international students (Erasmus + and others). The lectures and seminars will be held in English language.

There are no additional conditions for visiting students, who wish to enrol to this elective course.

Additional explications and notes:

1. The Course Regime enters in force on the date of issue and remains valid until its revocation or alteration. The Course Regime may not be altered during the academic year. Any changes to the Course Regime may only enter into force starting with the next academic year (changes must be submitted no later than 14 days prior to the start of the academic year as the **new Course Regime**).
2. The Course Regime for *compulsory courses* must be published no later than 14 days prior to the start of the academic year.
3. The Course Regime for *elective courses* is also considered the Elective Course Announcement and must be published no later than the 30th of July prior to the start of the academic year in which it enters in force.
4. The Course Regime must be published in Slovenian and English.
5. In the Course Regime, the »Regulations for the Assessment of Knowledge and Skills for the Single-Cycle Master Study Programmes Medicine and Dental Medicine« will be referred to as "*Regulations*".

In case of any further questions, please contact the Study Affairs Commission of the UL MF, via e-mail: ksz@mf.uni-lj.si.