

Semmelweis University, Faculty of Medicine - single, long-cycle medical training**Name of the host institution (and any contributing institution):**

Bioinformatika Tanszék

Name of subject: Klinikai bioinformatika**in English:** Clinical bioinformatics**in German:** Klinische Bioinformatics**Credit value:** 3**Semester:** 5. szemeszter

(in which the subject is taught according to the curriculum)

Hours per week	Lecture	Practical lesson	Seminar
0.0	0.0	0.0	0.0

Hours per semester	Lecture	Practical lesson	Seminar
43.0	19.0	24.0	0.0

Type of course:

optional

Academic year:

2025/26

Language of instruction (for optional and elective subjects):

english

Course code:

AOVBIN1238_1A

(in the case of a new course, to be completed by the Dean's Office, following approval)

Course coordinator name: Dr. Gyórfy Balázs**Course coordinator location of work, telephone availability:** Bioinformatika Tanszék,
06-30-514-2822**Course coordinator position:**tanszékvezető egyetemi tanár**Course coordinator Date and number of habilitation:**2017.03.21., PPKE-J-1/2017 Hab**Objective of instruction and its place in the curriculum:**

Two-thirds of recent biomedical research publications include a bioinformatics tool. What are they? During the course, four primary areas of bioinformatics (fundamentals of bioinformatics, omics, integrative science, and artificial intelligence) will be presented in lectures. During

the practical sessions, students will gain hands-on experience with cutting-edge software for data analysis, including a detailed evaluation of next-generation sequencing data. During the course, students will acquire valuable practical knowledge in everyday scientific work in the future (e.g., TDK, thesis, Ph.D., clinical research).

Method of instruction (lecture, group work, practical lesson, etc.):

theory (58 15-minute lectures) + practice (24 45-minute exercises)

Competencies acquired through completion of course:

The student will become familiar with the available bioinformatics methods and will be able to use them

The disadvantages and advantages of each method, the results of research and the interpretation of research results and clinical trials.

Course outcome (names and codes of related subjects):

Prerequisites for course registration and completion: (CODE):

Successful first 4 semesters (Medical Physiology II, Molecular Cell Biology II)

In the case of multi-semester courses, position on the possibility of and conditions for concurrent registration:

-

The number of students required to start the course (minimum, maximum), student selection method:

Minimum number of students: 20

Maximum number of students: 60

Detailed course syllabus (if the course can be divided into modules, please indicate): (Theoretical and practical instruction must be broken down into hours (weeks), numbered separately; names of instructors and lecturers must be listed, indicating guest lecturers/instructors. It cannot be attached separately! For guest lecturers, attachment of CV is required in all cases!)

The topics given in the thematic guide are 15-minute lectures.

Theory:

I. block: Basics of Bioinformatics

- A1. Introduction to bioinformatics (Dr. Balázs Gyórfy)
- A2. Utilization of a training and test set (Dr. János Tibor Fekete)
- A3. Statistical errors and dichotomania (Dr. János Tibor Fekete)
- A4. Survival analysis: Cox regression and the Kaplan-Meier plot (Prof. Dr. Balázs Gyórfy)
- A5. ROC analysis: predicting sensitivity and specificity (Dr. János Tibor Fekete)
- A6. Multiple hypothesis testing (Prof. Dr. Balázs Gyórfy)
- A7. Regression methods (Dr. Otília Menyhárt)
- A8. Meta-analysis: Visualization tools (Prof. Dr. Balázs Gyórfy)
- A9. Meta-analysis: statistical methods (Dr. János Tibor Fekete)
- A10. Network meta-analysis (Dr. János Tibor Fekete)

Block I theoretical hours: 3,3 hours (150 minutes)

II. block: Genomics

- G1. Introduction to genomics (Prof. Dr. Balázs Gyórfy)
- G2. Similar genes and proteins, BLAST (Prof. Dr. Balázs Gyórfy)
- G3. Genomics: quality control (Dr. Gyöngyi Munkácsy)
- G4. Genomics: alignment of data to a reference genome (Dr. Gyöngyi Munkácsy)
- G5. Genomics: identifying mutations (SNV, indels) in normal and tumor samples (Dr. Gyöngyi Munkácsy)
- G6. Genomics: determining the consequence of a mutation (Dr. Gyöngyi Munkácsy)
- G7. Genomics: what is the clinical relevance of a mutation, ClinVar, dbSNP (Dr. Gyöngyi Munkácsy)
- G8. Genomics: copy number variations (Dr. Gyöngyi Munkácsy)
- G9. Genomics: identifying processing artefacts and quality issues (Dr. Otília Menyhárt)

Block II theoretical hours: 3 hours (135 minutes)

III. block: Transcriptomics

- T1. Proteomics and transcriptomics: pre-processing (Prof. Dr. Balázs Gyórfy)
- T2. Transcriptomics: Processing RNAseq data (Prof. Dr. Balázs Gyórfy)
- T3. Processing single-cell data (Prof. Dr. Balázs Gyórfy)
- T4. Correlation (Dr. Otília Menyhárt)
- T5. Graphical comparison of two groups: boxplot, violin plot, density plot, heatmap, correlation, matrix (Dr. Áron Bartha)
- T6. Basics of regression (Dr. Otília Menyhárt)
- T7. Gene ontology (Dr. János Tibor Fekete)

Block III theoretical hours: 2,3 hours (105 minutes)

IV. block: Proteomics

- P1. Antibody based evaluation (Dr. Gyöngyi Munkácsy)
- P2. Mass Spectrometry I - Qualitative Analysis of Proteins (Dr. Áron Bartha)

- P3. Mass Spectrometry II - Quantitative Measurement of Proteins (Dr. Áron Bartha)
- P4. Protein structure determination: physical/chemical methods (Dr. Gábor Tusnády)
- P5. Modelling protein structure: from simple (homology model) to complex (ab initio) (Dr. Gábor Tusnády)
- P6. Modelling the structure of transmembrane proteins (Dr. Gábor Tusnády)
- P7. Determination of biological functions and UniProt (Dr. László Dobson)
- P8. Identification of the functions of intrinsically disordered proteins (Dr. László Dobson)
- P9. Identification of functional protein modules (Dr. László Dobson)
- P10. Using Alpha Fold (Dr. László Dobson)

Block IV theoretical hours: 3.3 hours (150 minutes)

V. block: Artificial intelligence

- M1. Machine learning (Dr. János Tibor Fekete)
- M2. The Bayes rule (Dr. János Tibor Fekete)
- M3. Principal component analysis (Dr. Áron Bartha)
- M4. Determining distance (Dr. Máté Posta)
- M5. Clustering (Dr. Máté Posta)
- M6. Basics of neural network (Dr. Máté Posta)
- M7. Recursive neural network (Dr. Otília Menyhárt)
- M8. Convolutional neural network (Dr. László Dobson)
- M9. Clinical application of a decision tree (Dr. Áron Bartha)
- M10. Feature selection (Dr. Otília Menyhárt)
- M11. Support Vector Machines (Dr. János Tibor Fekete)
- M12. The general pre-trained transformer (Dr. János Tibor Fekete)

Block V theoretical hours: 4 hours (180 minutes)

VI. block: Integrative sciences

- X1. Multi-omics (Dr. Otília Menyhárt)
- X2. Reproducibility issues in medical research (Dr. Otília Menyhárt)
- X3. Evaluation of Chip-seq and ATAC-seq data (Dr. Bálint Bálint)
- X4. Evaluation of DNA methylation data (Dalma Müller)
- X5. Epigenetic databases (Dalma Müller)
- X6. Using Excel for database management (Dr. Gyöngyi Munkácsy)
- X7. Data mining and AI (Dr. Gyöngyi Munkácsy)
- X8. Using REDcap (Dr. Otília Menyhárt)
- X9. Time distortion and computer addiction (Dr. Otília Menyhárt)
- X10. Blockchain and data security (Dr. Áron Bartha)

Block VI theoretical hours: 3.3 hours (150 minutes)

Practical training:

I. blokk: Basics of Bioinformatics

GY1. Survival analysis: Cox regression and the Kaplan-Meier plot, 2 hours (Dr. Balázs Gyórfy)

GY2. ROC analysis, 2 hours (Dr. János Tibor Fekete)

GY3. Multiple hypothesis testing, 2 hours (Prof. Dr. Balázs Gyórfy)

GY4. Introduction to Galaxy, 1 hour (Dr. Otília Menyhárt)

Block I Practical hours: 7 hours (315 minutes)

II.-III blocks: Genomics and transcriptomics

GY5. Quality control, 2 hours (Dr. Gyöngyi Munkácsy)

GY6. Alignment of data to a reference genome, 2 hours (Dr. Gyöngyi Munkácsy)

GY7. Identifying variants, 2 hours (Szonja Anna Kovács)

GY8. Consequences of mutations, 2 hours (Dr. Gyöngyi Munkácsy)

GY9. Transcriptomics: processing RNA-seq data, 2 hours (Prof. Dr. Balázs Gyórfy)

Blocks II-III Practical hours: 10 hours (450 minutes)

IV. block : Artificial intelligence

GY10. Clustering, 1 hour (Dr. Otília Menyhárt)

GY11. Classification, 2 hours (Dr. János Tibor Fekete)

GY12. Regression, 1 hour, (Dr. Otília Menyhárt)

GY13. Deep learning, 2 hours (Dr. János Tibor Fekete)

Block V Practical hours: 6 hours (270 minutes)

VI. block: Integrative science

GY14. Epigenetics, 1 hour (Dr. Bálint Bálint)

Block VI. Practical hours: 1 hour (45 minutes)

A consultation is possible during the last practical session.

Total number of theoretical hours: 19.3 hours (870 minutes)

Total number of practical hours: 24 hours (1080 minutes)

Day	Theory covered	Practical topics	Total hours
1	A1-A8	GY1, GY2, GY3	8.7
2	A9-A10; G1-G9	GY4, GY5, GY6	8.7
3	T1-T7; P1	GY7, GY8, GY9	8.7
4	P2-P4; M1-M11	GY10, GY11, GY12	8.7
5	P5-P10; M12; X1-X10	GY13, GY14	8.7

Other courses with overlapping topics (obligatory, optional, or elective courses) in interdisciplinary areas. To minimize overlaps, topics should be coordinated. Code(s) of courses (to be provided):

Requirements for attendance, options for making up missed sessions, and method of absence justification:

In accordance with the current Study and Examination Regulations. Attendance at the exercises is compulsory and may be replaced by
may be made up in the course of the last practical session in the form of a consultation.

Assessment methods during semester (number, topics, and dates of midterms and reports, method of inclusion in the course grade, opportunities for make-up and improvement of marks):

(number, topics, and dates of midterms and reports, method of inclusion in the course grade, opportunities for make-up and improvement of marks)

Completion of the tasks described in the practical training, which accounts for 33.3% of the end-of-semester assessment.

Number and type of individual assignments to be completed, submission deadlines:

-

Requirements for the successful completion of the course:

The number of absences from lectures and tutorials should not exceed the number of days per semester

25% of the exercises of the semester.

Type of assessment:

projektfeladat_en

Examination requirements (list of examination topics, subject areas of tests, lists of mandatory parameters, figures, concepts and calculations, practical skills, optional topics for the project assignment recognized as an exam and the criteria for its completion and evaluation)

The project task consists of a bioinformatics exercise, which are identical to one of the practical exercises studied during the semester, but which contain new data.

The bioinformatics task can be performed using any assistive device, including the use of your own laptop.

Method and type of grading (Share of theoretical and practical examinations in the overall evaluation. Inclusion of the results in the end-of-term assessment. Possibilities of and conditions for offered grades.): (Share of theoretical and practical examinations in the overall evaluation, Inclusion of the results in the end-of-term assessment, Possibilities of and conditions for offered grades)

The end-of-semester grade is based on the tasks described in the practical training (33.3% if

minimum 75% of the answers are submitted) and the project assignment (67.7%).

Signature of habilitated instructor (course coordinator) announcing the course:

Signature of the director of the host institution:

Date of submission:
