COURSE DESCRIPTION CARD - SYLLABUS

Course name
Structural Variation of Genomes [S2Bioinf2>ZSG]

Course

Field of study
Bioinformatics

Year/Semester
2/3

Area of study (specialization)
–

Profile of study
general academic

Level of study
second-cycle

Course offered in
Polish

Form of study
full-time

Requirements
elective

Number of hours

Lecture
30

Laboratory classes
30

Other (e.g. online)
0

Tutorials
0

Projects/seminars
0

Number of credit points

4,00

Coordinators

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Lecturers

Prerequisites

The person undertaking studies on the second degree of the Bioinformatics should have achieved effects of educating this subject from the first degree, defined in a PUT senate resolution - these effects are being presented in online services of the faculty http://www.cat.put.poznan.pl/. The student should have a basic knowledge in the molecular biology, genomics and transcriptomics, as well as of the knowledge of programming, statistics and biocomputer analysis of biological sequences. He/she should possess a skill of solving biological and bioinformatic basic problems, testing and correcting errors in implement programs, and skills of acquiring information from indicated sources and using databases. Moreover, in social competence the student must present such attitudes, like honesty, responsibility, perseverance, cognitive curiosity, creativity, propriety, respect for other people.

Course objective

1. Presentation of the basic knowledge to students about different kinds of structural variations of genomes, their influence on diversifying phenotypes and meaning for biology, biotechnology and medicine 2. Presentation of different analytical approaches in the identification of structural variants (in particular of the duplication and the deletion) in eukaryotic genomes based on high-thruoghput technologies, showing how different stages of the analysis can affect results 3. Acquainting students with chosen databases with data on the structural variation of genomes, recording formats and manners of visualisations of this data 4. Developing the ability of understanding the scientific literature, the reading and discussion with students

Course-related learning outcomes

Knowledge:
1. has a knowledge, with theoretical basis in bioinformatics approaches in analysis of the structural variation of genomes
2. knows detailed issues of bioinformatics analysis in the genome scale and on the level of the population based on solid theoretical bases
3. knows methods, techniques and tools used in the process of solving bioinformatics complex tasks, mainly of engineering character
4. knows principles of planning reasearch in bioinformatics field

Skills:
1. is able to obtain source data described in academic publications (e.g. results of genomic analyses), to acquire from various sources and to interpret information to their subject (e.g. functional annotations, paths of connections)
2. is able to show bioinformatics appropriate methods for the identification and genotyping of structural variants and to put them into practice
3. is able to plan and to build the pipeline of the data analysis from the sequencing of genomes under the angle of analysis of the structural variation, including the specificity of data (way of sequencing, kind, ploidity and similair).
4. knows and applies statistical methods in data submitted to genomic analysis and of identification of meaningful results
5. is preparing the presentation of results of research works in Polish and English, is discussing obtained results basing on the existing scientific knowledge
6. is formulating and testing hypotheses associated with bioinformatics problems.

Social competences:
1. Develops an attitude of openness to cooperation in the academic and industrial environment, working to respect the principles of ethics and good practices in the analysis of biological and biomedical data.
2. Is aware of the importance of protecting sensitive data and complies with the principles related to their processing, in particular in the context of genomic data, which may have a direct impact on the health and privacy of patients.

Methods for verifying learning outcomes and assessment criteria

Learning outcomes presented above are verified as follows:

Forming evaluation
a) in lectures verifying established effects of the education is being carried out through:
• the activity in discussion to the subject discussed;
b) in laboratories verifying established effects of the education is being carried out through:
• on the basis of assessment of current progress of the execution of tasks;
Evaluation
a) in lectures verifying established effects of the education is being carried out through:
• filing the written test with 5 questions - every task 0-4 pt (tasks can consist of a few subsections - there is a then set fragmentary score for every subsection). In order to get the credit one should score at least 11 points. In case of absence on more than 1/3 of the lectures will require additionally of writting a review of on of the particles in the scope of lectures
b) in laboratories:
the final grade is the average of the grade for the development and presentation of a selected tool or scientific publication and the grades for the performance of individual practical exercises in the field of

analysis of structural variability of genomes during the semester. A maximum of 10 points can be awarded for the presentation of a selected tool or publication. A maximum of 5 points can be awarded for each exercise/report (8-10 exercises). A pass can be obtained after obtaining more than 50% of the total number of points, provided that all required reports are submitted.

Programme content

New sequencing technologies created the possibility of the sequencing and comparing of individual genomes representing various populations and cataloguing the genetic variation both to the scale of single nucleotides as well as great structural variants. Based on such data it is possible to inspect genetic bases of evolutionary, adaptive or pathological processes. The object is devoted to presenting kinds and causes of structural variation of genomes, molecular mechanisms of their coming into existence, influence of the variation on diversifying phenotypes and their meaning for biology, biotechnology and medicine. Selected programs and methods of analyses will be introduced to the structural variation based on omics data and existing repositories and international initiatives of cataloguing the scope of the structural variation of the human and model organisms. Issues will be illustrated with examples from the scientific literature. Laboratories consist in the work with presented programs presented during lectures and analyses of biological and biomedical data for structural variatin in population. Moreover students are choosing the articles devoted to the scope of the lecture and present the problems decribed there.

Course topics

Discussion of the genetic basis of evolutionary processes.
Causes and types of genome variation in population studies.
GWAS - an approach used in genetic studies to link specific genetic variants to specific diseases.
Rare diseases - how are they detected.

Teaching methods

Multimedia presentation, discussion, group work, practical excerices

Bibliography

Basic:
Article, tutorials and manuals

Additional:
Hartl, Clark „Podstawy genetyki populacyjnej” Wydawnictwa Uniwersytetu Warszawskiego 2009, Wyd. I

Breakdown of average student's workload

	Hours	ECTS
Total workload	105	4,00
Classes requiring direct contact with the teacher	60	2,50
Student's own work (literature studies, preparation for laboratory classes/ tutorials, preparation for tests/exam, project preparation)	45	1,50