



## COURSE DESCRIPTION CARD - SYLLABUS

Course name

Structural Variation of Genomes

### Course

Field of study

Bioinformatics

Area of study (specialization)

Level of study

Second-cycle studies

Form of study

full-time

Year/Semester

2/3

Profile of study

general academic

Course offered in

Polish

Requirements

elective

### Number of hours

Lecture

30

Laboratory classes

30

Other (e.g. online)

Tutorials

Projects/seminars

### Number of credit points

4

### Lecturers

Responsible for the course/lecturer:

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Responsible for the course/lecturer:

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### 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-throughput 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 research 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, ploidy and similar).
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. is able to cooperate and to work in the group, taking different roles on in it.
2. is able appropriately to determine priorities serving the performance of a task determined by oneself or other.
3. is showing the creative posture in the working life and social.

### 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 5-7 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 writing a review of on of the particles in the scope of lectures

b) in laboratories:

The final evaluation is determined on the average evaluations of the exercises, practical laboratories and discussions about the scientific articles

### 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 variation in population. Moreover students are choosing the articles devoted to the scope of the lecture and present the problems described there.

### Teaching methods

Multimedia presentation, discussion, group work, practical exercises

### 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	100	4,0
Classes requiring direct contact with the teacher	60	2,0
Student's own work (literature studies, preparation for laboratory classes/tutorials, preparation for tests/exam, project preparation) <sup>1</sup>	40	2,0

<sup>1</sup> delete or add other activities as appropriate