

SYLLABUS

Name: Next Generation Sequencing (BioAu>SM2NGS20)

Name in Polish:

Name in English: Next Generation Sequencing

Information on course:

Course offered by department: Faculty of Automatic Control, Electronics and Computer Science

Course for department: Silesian University of Technology

Default type of course examination report:

ZAL

Language:

English

Course homepage:

<https://platforma2.polsl.pl/rau1/course/view.php?id=508>

Short description:

The objective of the course is to teach students the knowledge and skills necessary for analysis of data from high-throughput sequencing experiments. Lectures will provide an overview of NGS technologies and statistical preprocessing and analysis methods. The laboratories will consist of practical sessions for DNA-seq data processing.

Description:

Teaching modes and hours

ECTS:3

Total workload: 90 (45 contact hours / 45 student's own work hours)

Lecture 15h

Laboratory 30h

Student's own work: preparation for classes, preparation of reports

Total workload required to achieve learning outcomes (contact hours / Student workload hours):

Lecture 15/15

Laboratory 30/30

Workload sum: 90

Lectures:

- 1) NGS biological background and mechanisms
- 2) Next Generation Sequencing technologies (Illumina, Roche 454, Ion Torrent, ABI SOLiD, PacBio, Oxford Nanopore) and file formats
- 4) DNA-seq data analysis - quality assessment, preprocessing and downstream analysis tools and methods: trimming, error correction, de-duplication, normalization, alignment and refinement, variant calling, de novo assembly
- 5) RNA-seq data analysis - preprocessing and downstream analysis tools and methods: alignment, expression level extraction, differential expression analysis, functional analysis
- 6) Single cell sequencing data analysis - experimental design and downstream analysis tools and methods: normalization of cell specific bias, computing size factors, highly variable gene identification

Laboratory:

- 1) Introduction to the Linux system environment and command line
- 2) Data quality assessment - FastQC
- 3) Data preprocessing I - alignment (bwa, tophat, STAR)
- 4) Data preprocessing II - refinement (Trimmomatic, SOAPec)
- 5) Data visualization - IGV
- 6) DNA-seq analysis
- 7) RNA-seq analysis
- 8) Single cell RNA-seq analysis

Bibliography:

- Mardis ER. Next-generation DNA sequencing methods. *Annu Rev Genomics Hum Genet.* 2008;9:387-402
- Wang, Xinkun. Next-generation sequencing data analysis. CRC Press, 2016.
- Goodwin, Sara, John D. McPherson, and W. Richard McCombie. "Coming of age: ten years of next-generation sequencing technologies." *Nature Reviews Genetics* 17.6 (2016): 333.
- Metzker, Michael L. "Sequencing technologies—the next generation." *Nature reviews genetics* 11.1 (2010): 31.
- Trapnell, C., Pachter, L. & Salzberg, S. L. TopHat: discovering splice junctions with RNA-Seq. *Bioinformatics* 25, 1105–1111 (2009).
- Trapnell, C. et al. Transcript assembly and quantification by RNA-Seq reveals unannotated transcripts and isoform switching during cell differentiation. *Nat. Biotechnol.* 28, 511–515 (2010).
- Langmead, B., Trapnell, C., Pop, M. & Salzberg, S. L. Ultrafast and memory-efficient alignment of short DNA sequences to the human genome. *Genome Biol.* 10, R25 (2009).
- Roberts, A., Pimentel, H., Trapnell, C. & Pachter, L. Identification of novel transcripts in annotated genomes using RNA-Seq. *Bioinformatics* 27, 2325–2329 (2011).
- Roberts, A., Trapnell, C., Donaghey, J., Rinn, J. L. & Pachter, L. Improving RNA-Seq expression estimates by correcting for fragment bias. *Genome Biol.* 12, R22 (2011).
- Love MI, Huber W and Anders S. Moderated estimation of fold change and dispersion for RNA-seq data with DESeq2. *Genome Biology*, 15, pp. 550 (2014).
- Young MD, Wakefield MJ, Smyth GK and Oshlack A. "Gene ontology analysis for RNA-seq: accounting for selection bias." *Genome Biology*, 11, pp. R14 (2010).
- Anders S, Reyes A and Huber W. Detecting differential usage of exons from RNA-seq data. *Genome Research*, 22, pp. 4025 (2012).

USOS: Szczegóły przedmiotu: BioAu>SM2NGS20, w cyklu: <brak>, jednostka dawcy: <brak>, grupa przedm.: <brak>

Dobin A. et al. STAR: ultrafast universal RNA-seq aligner. Bioinformatics 29. 15-21 (2012).

Learning outcomes:

The student knows different technologies of NGS; K2A_W04, K2A_W09, K2A_W10, K2A_W11, K2A_K01

The student knows basic concepts of molecular biology and high-throughput sequencing, understands the differences between NGS applications (ex. RNA-seq, DNA-seq, WES, WGS, scRNA-seq); K2A_W04, K2A_W09, K2A_W11, K2A_K01

The student knows the analysis pipeline and is familiar with statistical methods used for the analysis; K2A_W17, K2A_W19, K2A_U10, K2A_K01

The student knows basic Unix commands and knows how to use software for NGS analysis; K2A_W11, K2A_U26, K2A_K01

The student can perform quality control of the data and perform proper data pre-processing, including adapter removal, BAM alignment and refinement. K2A_W11, K2A_W17, K2A_W19, K2A_U10, K2A_K01

The student is able to analyze the aligned data, infer and make decisions about the appropriate downstream analysis; K2A_W17, K2A_W19, K2A_K01, K2A_K02, K2A_K03

The student can efficiently use English literature sources; K2A_U01, K2A_U02, K2A_U06, K2A_K01

The student is able to collaborate in a group, can act creatively and make independent decisions about the choice of best analysis pipeline; K2A_U10, K2A_K01, K2A_K02, K2A_K03, K2A_K06

Assessment methods and assessment criteria:

Laboratory assignment reports.

Passing criteria: minimum 40% in every laboratory topic

The syllabus is valid from the winter semester / academic year 2025/2026 and its content is not subject to change during the semester

Course credits in various terms:

<without a specific program>

Type of credits	Number	First term	Last term
European Credit Transfer System (ECTS)	3	2021/2022-Z	