

INSTITUTE OF BIOMEDICINE

BMA231 Next Generation Sequencing Data Analysis with clinical applications, 7.5 credits

Next Generation Sequencing dataanalys med kliniska tillämpningar, 7,5 högskolepoäng Second Cycle

Confirmation

This course syllabus was confirmed by Institute of Biomedicine on 2023-02-01 to be valid from 2023-08-28, autumn semester of 2023.

Field of education: Medicine 100% *Department:* Institute of Biomedicine

Position in the educational system

The course is given as a stand-alone course. The course may be part of the master's / master's degree in biomedical laboratory science.

Main field of studies	Specialization
Biomedical Laboratory Science	A1N, Second cycle, has only first-cycle
	course/s as entry requirements

Entry requirements

For admission to the course, a Bachelor's degree is required of 180 credits or equivalent in any of the fields of medicine, nursing or natural sciences and English 6.

Learning outcomes

On successful completion of the course the student will be able to:

Knowledge and understanding

• Identify the different "Next Generation Sequencing" technologies that are currently available

• Describe the workflow a sample undergoes from library preparation to raw sequencing data

• Categorize "Next Generation Sequencing"-datasets according to standard quality thresholds

• List bioinformatics tools used in the analysis of "Next Generation Sequencing"-data

• Describe the typical workflow in the analysis of targeted resequencing and RNA-sequencing data

Competence and skills

• Propose a suitable experiment design in the analysis of "Next Generation Sequencing"data

- Analyze and interpret targeted resequencing and RNA sequencing data
- Modify the common "Next Generation Sequencing"-workflows to suit different cases
- Use fundamental commands in R for basic data manipulation and plotting
- Use and, when needed, modify existing R scripts

Judgement and approach

• Review the in silico techniques that have been introduced in the course

• Assess the quality of a sequencing project

• Select a specific "Next Generation Sequencing"-workflow based on the sequencing project design

• Evaluate the significance of their own and others' scientific results

Course content

In this course, students will focus on the analysis and interpretation of clinical "Next Generation Sequencing" (NGS) data applying various bioinformatics webtools. The statistical tool R will be introduced to perform and visualize the results. The course covers essential concepts in molecular biology and genetics as well as the principles on "Next Generation Sequencing"-applications, with a focus on targeted resequencing and RNA-sequencing. Students will perform in depth analysis of "Next Generation Sequencing"-quality assessment and interpretation of mutation and gene expression analyses.

Form of teaching

The course includes a combination of lectures, seminars and written assignments.

The course is given remotely.

Language of instruction: English

Assessment

The examination consists of:

- 8 written assignments
- A written final report presenting the results from the analysis of (publicly
- available) clinical datasets
- An oral presentation of the final report.

In addition to the above, it is mandatory for the student to attend 6 seminars. Any absence from a seminar is covered by a compensatory assignment.

Any absence from the oral presentation will be made up at a later given time.

If a student who has failed twice in the same examination section wishes to change the examiner before the next examination session, such a request should be submitted in writing to the department and must be approved unless there are special reasons to the contrary (HF ch. $6, \S 22$)

If the student has received a recommendation from the University of Gothenburg for special support, the examiner can, if it is compatible with the course's goals and provided that unreasonable resources are not required, decide to give the student an adapted examination or an alternative form of examination.

In the event that a course has ceased or undergone a major change, the student must be guaranteed at least three examination opportunities (including regular examination opportunities) for a period of at least one year, but no longer than two years after the course has ceased/changed. With regard to practice and work-based education, only one additional examination opportunity applies.

Grades

The grading scale comprises: Pass (G) and Fail (U).

To receive a Pass (G) on the course, the student is required to complete and pass (G) all Ladok modules.

Course evaluation

Course evaluation is made in writing with help of a general course evaluation, as well as orally in dialogue with the students. The course cordinator summarises and makes an analysis of the course evaluation and provides suggestions for development of the course. Analysis and suggestions are reconnected to the students and published on the Gothenburg University's Learning management platform, and presented at the next start of the course.

Additional information

As the course is given remotely, and all communication regarding the course will take place via the university's learning platform, it is a requirement that the student has access to a computer and internet connection. Lectures, exercises, literature and other relevant information will also be posted on the learning platform.