



**Karolinska
Institutet**

Course syllabus for

Next generation sequencing (NGS) and bioinformatics, 7.5 credits

Next generation sequencing (NGS) och bioinformatik, 7.5 hp

This course syllabus is valid from autumn 2024.

Please note that the course syllabus is available in the following versions:

Autumn2024 , Autumn2025

Course code	3BL008
Course name	Next generation sequencing (NGS) and bioinformatics
Credits	7.5 credits
Form of Education	Higher Education, study regulation 2007
Main field of study	Biomedical Laboratory Science
Level	AV - Second cycle
Grading scale	Pass with distinction, Pass, Fail
Department	Department of Laboratory Medicine
Decided by	Education committee LABMED
Decision date	2024-03-21
Course syllabus valid from	Autumn 2024

Specific entry requirements

Completed biomedical laboratory science education and Degree of Bachelor of Science in Biomedical Laboratory Science of 180 credits or Bachelor's degree in biomedical laboratory science. In addition, at least passed degree in Swedish and English equivalent to Swedish B/Swedish 3 and English A/English 6.

Objectives

The general aim of the course is that the students should develop advanced knowledge about and understanding of next generation sequencing (NGS) and bioinformatic analysis in genomics and transcriptomics. The aim is also to give an insight in how the methods are used in clinical diagnostics and other fields relevant for biomedical scientists.

On completion of the course, the student should be able to:

Knowledge and understanding

- Compare and explain the molecular principles behind NGS and have an overview of the methods of analysis that are used for NGS data.

- Relate different applications of NGS to clinical issues and research hypotheses and reflect on possibilities and challenges.
- Reflect on e.g. patient-ethical, integrity and data security related, societal and economical issues associated with these methods in clinical activities.

Skills and abilities

- Apply different applications based on NGS, analyse data from NGS and critically evaluate result.
- Independently assess the suitability of several applications based on NGS for different purposes.
- Reflect and theorise around different analyses based on NGS, and in writing compile material and orally give an account of its applications in diagnostics and research.
- Reflect on the development of NGS and bioinformatics and their importance for diagnostics of diseases and research with perspectives on societal issues.
- Problematising over ethical issues around NGS data.

Evaluation skills and way of approach

- Apply a scientific and reflecting approach.

Content

The course content is based on and is a specialisation of previous knowledge in molecular diagnostics in the undergraduate programme for biomedical scientists. The course is divided into different themes, where the students acquire advanced knowledge according to below:

- The methodology behind NGS - Massive parallel sequencing.
- Different applications of NGS such as DNA-sequencing, RNA sequencing, epigenetics, metagenomics, transcriptomics.
- Bioinformatic analysis of various types of NGS-data.
- Applications of NGS in diagnostics and research with e.g. mutation analyses and transcription analyses of e.g. cancer, microbiomes and viruses.
- Patient-ethical, integrity and data security related, societal and economical issues.

Teaching methods

The pedagogy is based on student-centered and student-activated learning.

Each session begins with students engaging with teacher-prepared materials such as video presentations and scientific articles. Afterwards, students meet digitally with teachers for discussions on the topic and for demonstrations such as bioinformatic data analyses. Additionally, students complete a written assignment.

Examination

A Pass grade requires active participation in teacher-supervised activities and a passed written examination which is graded Fail/Pass/Pass with distinction.

The teacher-supervised activities and the project presentations are compulsory. The examiner decides if, and how, absence from compulsory parts can be compensated. Study results cannot be reported until the student has participated in compulsory course elements or compensated for any absence in accordance with instructions from the examiner. Absence from a compulsory educational component may mean that the student cannot take the exam until the next time the course is given.

Students who do not pass a regular examination are entitled to re-examination on five more occasions. If the student has carried out six failed examinations/tests, no additional examination or new course

admission is approved.

Each occasion the student participates in the same test counts as an examination. Submission of a blank exam is considered an examination occasion. In case a student is registered for an examination but does not attend, this is not regarded as an examination.

In the event of special circumstances, or if a student with a disability is in need of certain adjustments, the examiner may decide to depart from the regulations on examination form, number of examination opportunities, possibility of completion or exemption from compulsory educational elements, etc. Content and intended learning outcomes as well as the level of expected skills, knowledge and abilities must not be altered, removed or lowered.

Other directives

The course is given in Swedish while certain parts, and course material are given in English. Course evaluation is carried out according to the guidelines that are established by the Committee for higher education

Literature and other teaching aids