Course syllabus for

Sequencing and Genomics in Diagnostics and Personalized Medicine, 4.5 credits

Sekvensering och genomik inom diagnostik och personaliserad medicin, 4.5 hp
This course syllabus is valid from autumn 2024.

Course code	4BI132
Course name	Sequencing and Genomics in Diagnostics and Personalized Medicine
Credits	4.5 credits
Form of Education	Higher Education, study regulation 2007
Main field of study	Biomedicine
Level	AV - Second cycle
Grading scale	Fail (U) or pass (G)
Department	Department of Medicine, Huddinge
Decided by	Programme committee for study programmes in biomedicine
Decision date	2024-03-11
Course syllabus valid from	Autumn 2024

Specific entry requirements

A Bachelor´s degree or a professional degree worth at least 180 credits in biomedicine, biotechnology, cellular and molecular biology, medicine, or the equivalent. Proficiency in English equivalent to the Swedish upper secondary school course English 6/English B.

Objectives

The aims of the course are that the student should be able to explain and give examples of how different types of genomics profiles can be utilized in diagnostics and personalized medicine that is based on molecular subtyping of patients. Furthermore, the course provides training in the critical evaluation of sequencing data (especially whole genome and targeted DNA sequencing) by understanding key steps in the data generation, principles of data analysis tools, and challenges in prioritizing the DNA variants from the collected results.

On completion of the course, the student should be able to:
- understand how short-read sequencing data is generated and the basic quality control steps,
- explain how DNA variants can be analysed from deep sequencing data,
- understand the conceptual framework of Bayesian statistics
- use graphical-user-interface bioinformatics tools including genome-browsers and variant annotation tools,

- understand how bioinformatics workflows for genomics data are set up and benchmarked,
- understand results across multiple sequencing modalities and how such data collected across multiple cohorts can be accessed and utilized via multi-omics
  data portals.

Content

Overview of short read sequencing data generation. Detecting germline and somatic DNA sequence variants from deep sequencing data. Variant calling, annotation and prioritization. Statistical analysis of genotype-trait associations. Bioinformatics tools for genomics built around command-line tools. Benchmarking of methods and tools. Interpreting genome-wide data from patients across several measurement modalities. cBioportal for Cancer Genomics and working with multiomics patient data. Virtual practicals: hands-on exercises and demonstrations.

Teaching methods

The course is at Master's level, where students are assumed to be familiar with the most common study methods in higher education. The fundamental pedagogical view is based on learning as an active research process.

The learning and teaching activities include multiform learning, including video lectures on basic concepts and terminology, expert viewpoint articles/videos, exploring sequencing data and results of statistical comparisons in graphical user-interface tools and computational exercises with Jupyter notebooks. Online teaching platforms will be utilized during the course with students having the possibility to interact in digital group rooms during practical sessions.

This online course has a suggested time table for studies and students enrolling at the same time will be able to interact with each other during the course. The final assignment can be performed as a team (strongly suggested to increase interaction between learners). Teachers will monitor discussion forums on weekly basis to address questions, offer additional advice and highlight recent articles related to course topics that can be studied as extra material. Teachers are also present in indicated digital breakout sessions.

Examination

The examination is a written assignment and written evaluation of another student's assignment (peer review). Graded Pass or Fail.

Compulsory participation
This is an online course run from University of Eastern Finland, Kuopio and to complete the course the student must participate and perform in all teaching and learning activities. The course examiner assesses if and, in that case, how absence from compulsory components can be compensated for. A student's study results cannot be finalised/registered until the student has participated in the compulsory components or compensated for their absence in accordance with the examiner's instructions. Absence from a compulsory component may mean that the student cannot compensate for absence until the next time the course is given.

Limitations of the number of examinations or practical training sessions
Students who have not passed the regular examination are entitled to participate in five more examinations. If the student has failed six examinations/tests, no additional examination or new admission is provided.

The number of times that the student has participated in one and the same examination is regarded as an examination session. Submission of a blank examination is regarded as an examination. An examination for which the student registered but not participated in, will not be counted as an examination.

If there are special grounds, or a need for adaptation for a student with a disability, the examiner may

decide to deviate from the syllabus's regulations on the examination form, the number of examination opportunities, the possibility of supplementation or exemptions from the compulsory section/s of the course etc. Content and learning outcomes as well as the level of expected skills, knowledge and abilities may not be changed, removed or reduced.

Other directives

This course is run by Institute of Biomedicine, university of Eastern Finland in an online mode.

The course language is English and examination is performed in English.

Course evaluation will be carried out in accordance with the guidelines established by the Committee for Higher Education.

This course replaces the course Sequencing and Genomics in Diagnostics and Personalized Medicine, 4.5 credits (4BI112) and cannot be included in a degree together with the latter course.

Literature and other teaching aids

Course materials are hosted in a Moodle platform at university of Eastern Finland and include lecture videos, suggested reading in the form of original articles and reviews and links to publicly available webinars and tutorials on the topics addressed. Jupyter notebooks are shared via a server that can be accessed during the course (accounts will be created for participants upon enrolling to the course).