DEPARTMENT OF MEDICAL EPIDEMIOLOGY AND BIOSTATISTICS

C8F6039, Clinical Cancer Genomics, 3 credits (hec)

Klinisk cancergenomik, 3 högskolepoäng

Third-cycle level / Forskarnivå

Approval

This syllabus was approved by the The Committee for Doctoral Education on 2024-09-23, and is valid from spring semester 2025.

Responsible department

Department of Medical Epidemiology and Biostatistics, Faculty of Medicine

Prerequisite courses, or equivalent

We expect each course participant to have basic Linux/Unix skills and to be able to run commands (programs) using a command-line interpreter or a shell terminal as available in Unix-like operating systems.

If you don't currently know how to do this, but would like to take part in the course, we recommend checking out online resources such as https://scicomp.aalto.fi/scicomp/shell/ or this two-hour online introduction: https://www.hpc2n.umu.se/events/courses/2024/fall/intro-linux.

Purpose & Intended learning outcomes

Purpose

This course aims to provide an introduction to cancer genomics and to support to obtain practical knowledge regarding how to apply state of the art methodology to interrogate the cancer genome in a routine clinical setting or a clinical trial setting. The course will include lectures covering the technology advancements that have enabled high-throughput analysis of cancer genomes and the knowledge that can be obtained by applying these technologies. This encompasses both laboratory sample processing and downstream bioinformatics and a mix of lectures and computer-based exercises. The exercises will include processing and analysis of DNA- and RNA-sequencing data covering file formats, quality control aspects, identification of somatic variation, curation of identified somatic- and germline variants for clinical use, clonality estimation and annotation of variants.

The main objective of the course is to facilitate that students get an understanding of basic theory and obtain practical knowledge that will enable course participants to apply the covered methodologies in their own research- or clinical laboratory.

Intended learning outcomes

At the end of this course the student will be able to:
• demonstrate a basic insight into the cancer genome.
• explain how the cancer genome can be interrogated through tissues and liquid biopsies.
• summarize how to apply technology to obtain relevant information from the cancer genome.
• summarize the file formats used in high throughput sequencing.
• apply command line bioinformatic tools for genomic analysis.
• summarize the constituents of a bioinformatics pipeline for processing Illumina sequencing data and to run such a pipeline.
• perform quality control on DNA- and RNA sequencing data for cancer sequencing purposes.
• call somatic- and germline variation.
• curate somatic- and germline variation.
• annotate somatic- and germline variation.
• use online resources such as genome browsers and portals for variant annotation.

Course content

• An introduction to the cancer genome and mutational processes in cancer.
• Overview of disease heterogeneity – the concept of cancer subtypes.
• The clinical impact of analysing the cancer genome.
• The concept of personalized therapy by tumour profiling.
• Intra-patient tumour heterogeneity.
• How to enable cancer genomics through tissues and liquid biopsies
• How to apply to high-throughput methodology to interrogate the cancer genome.
• Illumina sequencing file formats.
• Bioinformatics pipelines.
• Processing of DNA- and RNA sequencing data.
• QC of both DNA- and RNA sequencing data
• Calling somatic- and germline variation:
- Point mutations and indels.
- Copy-number alterations.
- Structural variation.
• File formats for variant calling.
• Annotating somatic- and germline variation.
• How to curate somatic- and germline variation for clinical use.

Forms of teaching and learning

A mix of lectures and computer exercises.

Language of instruction

The course is given in English

Grading scale

Pass (G) /Fail (U)

Compulsory components & forms of assessment

Compulsory components

All sessions are compulsory. Each session is exclusive and cannot be compensated for later on. The student will be asked to review the issue presented in case of absence from a session.

Forms of assessment

Each computer exercise addresses one or multiple learning outcomes. Each student will hand in a written report from each computer exercise. All intended learning outcomes need to be achieved in order to pass the course.

Course literature

Recommended reading before the course:
Clinical cancer genomic profiling https://doi.org/10.1038/s41576-021-00338-8.
Standard operating procedure for somatic variant refinement of sequencing data with paired tumor and normal samples https://doi.org/10.1038/s41436-018-0278-z