

Massively Parallel Sequencing Data-analysis (D012547)

Course size *(nominal values; actual values may depend on programme)*

Credits 3.0

Study time 75 h

Course offerings and teaching methods in academic year 2024-2025

A (semester 2)

English

Gent

seminar
lecture

Lecturers in academic year 2024-2025

Mestdagh, Pieter

GE31

lecturer-in-charge

Coppieters, Frauke

GE31

co-lecturer

Everaert, Celine

GE31

co-lecturer

Van Heetvelde, Mattias

GE31

co-lecturer

Offered in the following programmes in 2024-2025

[Master of Science in Biomedical Sciences](#)

crdts

3

offering

A

Teaching languages

English

Keywords

Massively parallel sequencing data analysis, RNA-sequencing, exome sequencing, targeted DNA sequencing, differential gene expression, alternative splicing, variants/mutations, single cell sequencing, R

Position of the course

The analysis of massively parallel sequencing (MPS) data is a crucial aspect of current molecular and genetic research. This involves both processing of raw data, analysis and visualization of results. To this end, dedicated pipelines (e.g. Galaxy) and advanced scripting (R) is required. The applications for RNA-seq will focus on differential gene expression analysis, splicing, fusion genes and variants, and for DNA-seq on gene copy numbers and variants (SNPs, mutations).

Contents

MPS workflow: processing, analysis and visualization

RNA-sequencing: read mapping (Tophat, Star), de novo transcript assembly, normalization and differential gene expression/splicing analysis (edgeR, DESeq, Limma Voom). Dealing with counts/RPKM/FPKM/TPM, pseudoaligners (Salmon, Sailfish, Kallisto), visualization of results (transcript structure, annotation, splicing (Sashimi)), single cell analysis (clustering, annotation and differential gene expression with Seurat).

DNA sequencing: Quality control of sequencing data (FASTQC), read mapping against the human genome and removal of duplicate reads (BWA, Picard), variant detection, local realignment (GATK), coverage analysis and read visualization (IGV). Variant filtering: integration of population frequencies (dbSNP, Exac, EVS), linkage data, patterns of inheritance, gene lists (including ACMG panel). Variant interpretation: predicting the functional effect of variants.

Initial competences

Successfully completed the courses on Statistics, Data Analysis I, Informatics II and Specialised Bio-informatics or having acquired the envisaged competences through an alternative track.

Successfully completed the bachelor training in Biomedical Sciences or having acquired the envisaged competences through an alternative track.

Final competences

- 1 Practical insights in the processing of massively parallel sequencing data
- 2 The capacity to successfully complete a similar analysis

Conditions for credit contract

Access to this course unit via a credit contract is determined after successful competences assessment

Conditions for exam contract

This course unit cannot be taken via an exam contract

Teaching methods

Seminar, Lecture

Extra information on the teaching methods

Courses and practical sessions

Study material

None

References**Course content-related study coaching**

Support during courses and practical sessions, support through Ufora

Assessment moments

end-of-term assessment

Examination methods in case of periodic assessment during the first examination period

Skills test, Written assessment with multiple-choice questions, Written assessment with open-ended questions

Examination methods in case of periodic assessment during the second examination period

Skills test, Written assessment with multiple-choice questions, Written assessment with open-ended questions

Examination methods in case of permanent assessment**Possibilities of retake in case of permanent assessment**

examination during the second examination period is possible in modified form

Extra information on the examination methods

Written examination on 50% of the total score (closed book) on matters listed in the course material or discussed during the course. Examination will be combination of multiple choice and open questions.
Practical examination (open book) on 50% of the total score.

Calculation of the examination mark

Written exam (50%) and practical exam (50%)