GHENT UNIVERSITY

Course Specifications

Valid as from the academic year 2025-2026

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 2025-2026

A (semester 2)	English	Gent	lecture
			seminar

Lecturers in academic year 2025-2026

Mestdagh, Pieter	GE31	lecturer-in-charge
Coppieters, Frauke	GE31	co-lecturer
Everaert, Celine	GE31	co-lecturer
Morlion, Annelien	GE31	co-lecturer
Van Heetvelde, Mattias	GE31	co-lecturer

Offered in the following programmes in 2025-2026	**crdts**	**offering**
Master of Science in Biomedical Sciences	3	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, Nextflow, reproducible bioinformatics analysis, high performance computing

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, Nextflow) 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 inherritance, gene lists (including ACMG panel). Variant interpretation: predicting the functional effect of variants.
Reproducible Bioinformatics Pipelines: running Nextflow pipelines adapted from NFCore and concepts for reproducible and large scale RNA and DNA-sequencing data analysis such as Docker containers and high performance computing.

Initial competences

Successfully completed the courses on Statistics, Data Analysis I, Informatics II and Specialised Bio-informatics or having aqcuired the envisaged competences through an alternative track.
Successfully completed the bachelor training in Biomedical Sciences or having aqcuired 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%). Moreover, for both parts a minimal score of 9/20 should be obtained to succeed for this course. In case for one part a score below 9/20 is obtained, the student will get a total score of maximum 9/20 (also in case the sum of both parts is above 9/20).