

# Course Specifications

Valid in the academic year 2024-2025

# Applied High-throughput Analysis (C003695)

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

Credits 6.0 Study time 180 h

# Course offerings and teaching methods in academic year 2024-2025

A (semester 1) English Gent seminar lecture

# Lecturers in academic year 2024-2025

De Meyer, Tim	LA26	lecturer-in-charge
Coussement, Louis	LA26	co-lecturer
Trypsteen, Wim	GE31	co-lecturer
Vandesompele, Jo	GE31	co-lecturer

Offered in the following programmes in 2024-2025	crdts	offering
Master of Science in Teaching in Science and Technology(main subject Biochemistry and Biotechnology)	6	Α
Bridging Programme Master of Science in Bioinformatics(main subject Engineering)	6	Α
Master of Science in Bioinformatics(main subject Bioscience Engineering)	6	Α
Master of Science in Bioinformatics(main subject Engineering)	6	Α
Master of Science in Bioinformatics(main subject Systems Biology)	6	Α
Master of Science in Biochemistry and Biotechnology	6	Α
Exchange programme in Biochemistry and Biotechnology (master's level)	6	Α
Exchange Programme in Bioinformatics (master's level)	6	Α

#### Teaching languages

English

# Keywords

omics data analysis, massive parallel sequencing, microarrays, kwantitatieve en digitale PCR, RNA-seq.

# Position of the course

This is an advanced course master of bioinformatics that focuses on the full data-analytical pipelines to (pre)process high throughput omics data. This will include the experimental protocols, as potential sources of variation/bias, image (microarray/sequencing) analysis leading to raw sequence/intensity data, quality control of the raw data, discussion and application of normalization and statistical analysis tools, quality control of data normalization/analysis and gene set/pathway analysis. The different steps will be described and illustrated by means of real examples in the areas of genomics, transcriptomics, and epigenomics. Importantly, the full data analytical pipeline is studied, focusing on conceptual insight rather than the theoretical details of the studied algorithms.

#### Contents

Each theoretical part will be followed by a practical session in which a data set will be analyzed making use of the state-of-the art tools.

Part 1: Introduction

- History and evolution of high throughput omics
- · Ethics of omics research, including scientific integrity and sustainability
- Omics data repositories and data sources (TCGA, GEO, ArrayExpress, SRA, ,...) Part 2: Microarrays
- Examples, quality control, applied data normalization and statistical analysis of:

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- expression microarrays (single and two-channel)
- comparative genomic hybridization arrays (CGH)
- SNP arrays / DNA methylation arrays
- Pathway / gene set analysis tools

Part 3: Quantitative sequencing

- Subtypes and experimental procedures: expression (RNA-seq), enrichment (ChIP-seq, MBD-seq/MethylCap-seq), bisulfite sequencing for DNA-methylation, metagenomics (species diversity)
- Raw data formats (e.g. FASTQ) and quality control
- Sequence alignment: standard mapping (e.g. BOWTIE), splice site aware mapping and isoform calling/quantification for RNA-seq (e.g. STAR, KALLISTO) and associated aligned data formats (e.g. SAM/BAM)
- Manipulation of large (sequencing) data sets (e.g. SAMTOOLS)
- Data summary using existing annotation (e.g. gene level, species level) or de novo (peak calling, particularly for ChIP-seq, e.g. MACS)
- Application of data normalization strategies (library size, trimmed mean of m-values method, etc. )
- Statistical analysis: identification of differential expression/enrichment (e.g. EdgeR, DESeq), modifications for single-cell transcriptomics.
- SNP and mutation calling, GWAS
- Pathway / gene set analysis tools

Part 4: High-throughput PCR based methods

- qPCR: quantification models, error propagation, normalization, experimental design, assay design
- dPCR:Poisson correction, advanced copy number analysis, power analysis, assay design

#### Initial competences

Basics molecular biology, statistical data analysis and scientific programming. Conceptual insight in general(ized) linear models is a plus.

#### Final competences

- 1 Understanding the application domains of high-throughput omics research, the generation of these data and the implications regarding bias and variance.
- 2 Knowledge of the different steps of the full omics analytical pipeline and how they are linked in a multidisciplinary fashion.
- 3 Be familiar with relevant omics databases, and its impact on scientific integrity and sustainability.
- 4 Have insight into the fast evolution of omics technologies and associated required bioinformatics solutions, and how this will/may have major implications for society for several important application domains such as medicine, food production and ecology.
- 5 Critical selection, evaluation and application of methods for high-throughput data (pre) processing.
- 6 Divide the omics analytical pipeline into separate steps and critically select the most appropriate methodology to solve each of these steps, taking into account both the practical applicability and the limitations/consequences of each methodology.
- 7 Report the outcome and particularities of a high-throughput omics analytical project to a non-specialist.
- 8 Apply fundamental knowledge of different subdomains (statistics, (bio) informatics) to plan and perform a complex data-analytical project at a professional level, taking into account financial, computational and other limitations.
- 9 Infer knowledge from complex high-throughput data using quantitative methods.10 To critically select and evaluate methods for high-throughput data-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

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#### Study material

Type: Syllabus

Name: Notes practical sessions

Indicative price: Free or paid by faculty

Optional: no
Language: English
Number of Pages: 200
Oldest Usable Edition: 2023
Available on Ufora: Yes
Online Available: No
Available in the Library: No

Available through Student Association: No

#### Type: Slides

Name: Slides AHTA

Indicative price: Free or paid by faculty

Optional: no
Language : English
Number of Slides : 500
Oldest Usable Edition : 2023
Available on Ufora : Yes
Online Available : No
Available in the Library : No

Available through Student Association: No

#### References

#### Course content-related study coaching

- Practica worden begeleid door assistenten.
- Bijkomende informatie via Ufora.

#### **Assessment moments**

end-of-term and continuous assessment

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

Oral assessment, Written assessment open-book

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

Oral assessment

# Examination methods in case of permanent assessment

Assignment

# Possibilities of retake in case of permanent assessment

examination during the second examination period is possible

## Extra information on the examination methods

- assignment: completion of a high-throughput omics data analytical project starting from several sources of raw data and communication of the results in a report for the non-specialist (e.g. clinician)
- open book examination: well defined problems associated with individual steps in the omics data analytical pipelines will be presented,
- oral examination: feedback regarding the assignment will be provided, and methodological aspects will be discussed

## Calculation of the examination mark

- Assignment report + discussion during oral examination: 50% of total mark
- Open book examination: 50% of total mark
- Student passes if final score for both parts combined is >=10/20

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