## Course Specifications

**Massively Parallel Sequencing Data-analysis (D012547)**

Valid as from the academic year 2018-2019

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### Course Offerings and Teaching Methods in Academic Year 2018-2019

<table>
<thead>
<tr>
<th>Course</th>
<th>Language</th>
<th>Type</th>
<th>Hours</th>
</tr>
</thead>
<tbody>
<tr>
<td>A (semester 2)</td>
<td>English</td>
<td>seminar</td>
<td>15.0 h</td>
</tr>
<tr>
<td></td>
<td></td>
<td>lecture</td>
<td>15.0 h</td>
</tr>
</tbody>
</table>

### Lecturers in Academic Year 2018-2019

- Mestdagh, Pieter GE02 lecturer-in-charge
- Coppieters, Frauke GE02 co-lecturer
- Lefever, Steve GE02 co-lecturer
- Rosseel, Toon GE02 co-lecturer

### Offered in the Following Programmes in 2018-2019

<table>
<thead>
<tr>
<th>Course</th>
<th>Credits</th>
<th>Offering</th>
</tr>
</thead>
<tbody>
<tr>
<td>Master of Science in Biomedical Sciences</td>
<td>3</td>
<td>A</td>
</tr>
</tbody>
</table>

### Teaching Languages

- English

### Keywords

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

### Position of the Course

The analysis of massively parallel sequencing 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 (i.e. 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.

### 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)).
  - DNA sequencing: Quality control of sequencing data (FASTQC), read mapping 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 (polyphen-2, SIFT, splice site prediction, Alamut Batch)

### Initial Competences

Successfully completed the courses on Statistics, Data Analysis I, Informatics II and advanced 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. Capability to successfully complete a similar analysis
3. Understanding literature on the subject

(Approved)
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
  Lecture, seminar, lecture: plenary exercises

Extra information on the teaching methods
  Courses and practical sessions

Learning materials and price
  English course, estimated cost: 15 euro

References

Course content-related study coaching
  Support during courses and practical sessions, support through Miverva

Evaluation methods
  end-of-term evaluation

Examination methods in case of periodic evaluation during the first examination period
  Written examination with open questions, written examination with multiple choice questions, skills test

Examination methods in case of periodic evaluation during the second examination period
  Written examination with open questions, written examination with multiple choice questions, skills test

Examination methods in case of permanent evaluation

Possibilities of retake in case of permanent evaluation
  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%)