

## Course

> Introduction to bioinformatic analysis of next (NGS) and 3rd generation sequencing data<

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<i>Lecturer</i>	<b>Coassin / Schönherr</b>
<i>Number</i>	049005
<i>Type / ECTS</i>	VU 2 / 1,5 ECTS
<i>Date/Time</i>	11.11.2019 – 14.11.2019
<i>Location</i>	PC training room 4 in Fritz-Preglstr. 3
<i>Limitations</i>	Min. of 4 participants; Max. of 8 participants
<i>Registration</i>	Register in i-med.inside Deadline 1 <sup>st</sup> of November 2019

## Description

Next generation sequencing methods are a pivotal technology in modern genetic research. While these technologies technically allow even a single scientist to generate sequence data of a full genome within days, the processing, management and interpretation of such an amount of data is challenging and heavily dependent on several data processing and sequence alignment algorithms. Therefore the steps between wet-lab library construction and the final result appear as a black box to many wet-lab oriented biologists. This is additionally worsened by the need to perform most steps in UNIX environment.

This course will focus on common short-read sequencing technologies (e.g. Illumina, IonTorrent) and also provide examples of Nanopore sequencing.

## Aim

This course provides an introduction on NGS technologies, NGS bioinformatics, resources for data analysis and variant annotation.

## Covered Topics

1. Introduction on the different second generation (Illumina, IonTorrent) and third generation sequencing (Pacific Bioscience, Nanopore Sequencing) technologies, library types and general steps of library preparation.
2. Steps of data processing
  - a. Introduction to UNIX and some useful commands (grep, awk, and others)
  - b. Concepts for handling large datasets
  - c. NGS data preprocessing
  - d. Sequence alignment and file formats SAM and BAM
  - e. Sequence variant calling (both germline and somatic mutations)
3. Sequence variant annotation, available reference data sets, genome and sequence databases for reference sequence identification and bioinformatic prediction of variant effects (possibilities and limitations).