Next Generation Sequencing Analysis

Next generation sequencing (NGS) methods continue to tremendously increase DNA sequence output, thereby greatly reducing the costs of genome-scale analyses. This has opened up a wide range of new sequencing applications aimed at understanding how species evolve and how cells function.

At the end of the course the students should have a detailed knowledge of bioinformatics methods for genome analysis using next-generation sequencing data. This includes knowledge of the existing types of sequencing data, how the different types of data can be displayed and analysed, the current methods for genome and transcriptome assembly and analysis, their accuracy, and how to apply them. The course will train the participants to devise and analyse a real genomics project involving NGS data. It will also train the student in presenting and communicating the results of an NGS analysis project in writing.

Learning Objectives
At the end of the course the students should be able to:
- **Describe** key challenges in analysis of NGS data.
- **Explain** the theoretical foundation of methods using NGS for assembling and analysing genomes.
- **Discuss** the bioinformatic methods for genome analysis.
- **Discuss** original literature within the subjects.
- **Use** bioinformatics tools within the selected application areas.

Contents/Topics
The course will start with an overview of current NGS technologies and directions in which these are currently progressing. Methods for assembling genomes from NGS data using either a reference genome or *de novo* assembly will be presented together with some emphasis on assessing NGS data quality (Quality control, accuracy of base calling). The course will then offer an overview of the range of biological/biomedical questions that these new types of data can help to address. This includes:
- How to build a Bioinformatics pipeline
- The use of NGS resequencing of closely related genomes for detecting mutations. This includes the characterization of somatic mutations in cancer tissues, the *de novo* mutation spectrum in eukaryotic genomes, and the detection of mutations underlying phenotypic changes in experimental or natural populations.
- Short-read (Illumina) versus long-read (Pacbio, Nanopore) sequencing
- The use of resequencing of a large number of individuals for Single Nucleotide Polymorphism (SNPs) and Copy number variation detection.
- Application of NGS in Transcriptomics and Epigenomics.
- Single-cell RNA-seq studies that go beyond tissue-type analysis to resolve single cell behavior
- Environmental/Meta-genomics studies probing the bacterial/viral diversity of understudied biomes.
- Comparative and evolutionary genomics.

To allow hands-on data analysis training, the course includes the following computer exercises, which can be completed using either your own or example data:
- Introduction to Galaxy, GATK and Samtools
- Grooming of reads
- Mapping of Illumina reads to reference
- SNP/indel calling, filtering and annotation
- *De novo* assembly of bacterial plasmids using short and long NGS reads.
- Analysis of single-cell RNA-seq data

Prerequisites
3 years study of molecular biology, biology, mathematics, engineering or computer science.

Types of Teaching
One week intensive course including lectures, colloquia and computer exercises.

Compulsory programme
Participation in the full one week course program.
Presentation of a mandatory NGS data analysis project report written as a scientific publication.

Lecturers
Mikkel Schierup and Stig U. Andersen.

Teaching Materials / Text books
Research and review papers from current scientific literature.

Course description
https://international.au.dk/education/admissions/summeruniversity/courses/next-generation-sequencing/

Evaluation
Individual written report

Credits
5 ECTS

Language
English

Capacity limit
none

Quarter
June 28th – July 2nd 2021

Examination periods
July 5th – July 9th 2021

Course Type
PhD course

Provider
Bioinformatics Research Center (BiRC)

Admission Requirements
See "Prerequisites"

Course Enrolment
Through AU Summer University
https://international.au.dk/education/admissions/summeruniversity/course/nextgenerationsequencing/ or by e-mail to teachers: mheide@birc.au.dk or sua@mbg.au.dk