## **Course Outline**

## 1. Course overview (1 lecture)

- a. Problems in computational biology including genome assembly in different flavours, RNA-seq, Chipseq, and other assays
- b. Single cell versions of various assays
- c. A discussion about the statistical and algorithmic challenges that are faced in these problems

## 2. High-throughput sequencing (1 lecture)

- a. Brief discussion of biological background
- b. Sequencing technologies (short read technologies like Illumina, long read technologies like Pacific Biosciences and Oxford Nanopore, linked read technologies like 10x)
- c. Base calling

#### 3. De novo Genome Assembly (3-4 lectures)

- a. Dense read formulation: Necessary and sufficient conditions (informational view)
- b. Algorithms for assembly: de Bruijn graph based algorithms, Overlap graph based algorithms
- c. Errors and biases

#### 4. Read alignment (3 lectures)

- a. Dynamic programming
- b. Hash-based seed-and-extend
- c. FM-index and Burrows-Wheeler transform
- d. Suffix arrays
- e. Minhash
- f. Applications such as spliced alignment, and alignments used in practical cases like DAligner, and Minimap.

## 5. Variant calling (1 lecture)

- a. SNV calling
- b. Structural variant calling

## 6. Phasing and Imputation (2 lectures)

- a. Imputation algorithms
- b. Phasing algorithms

#### 7. RNA-Seq assembly (2 lectures)

- a. Formulation
- b. Algorithms
- 8. RNA-Seq quantification (2 lectures)
  - a. EM algorithm
- 9. Single-cell RNA-Seq analysis (3 lectures)
  - a. Differential expression
  - b. Cell Differentiation
  - c. Visualisation
  - d. Trend Analysis
- 10. Genome Compression (1 lecture)

Guest lecture by Bikash Sabata, Vice President of Software at Genia, Roche Sequencing on 6 April 2016.

Guest lecture by Stephen Turner, Co-founder and Chief Technology Officer, Pacific Biosciences on 13 April 2016.

# **Useful Resources**

- Lawrence Hunter, Molecular Biology for Computer Scientists A crisp write-up on the basics of biology which motivate, and provide insights into problems we discuss in class. This is written in a non-biologist friendly manner.
- 2. Eric Lander, Fundamentals of Biology, MIT Open Course Ware- Lectures covering the basics of biology. Very friendly to non-biologists.
- 3. Ben Langmead's lecture notes Covers many topics that we cover in class. Some very nice video lectures and example code in ipython notebooks.
- 4. Bioinformatics algorithms by Compeau and Pevzner Covers many topics that we cover in this class. Video lectures are also available on the book site.