Course Outline

1. Overview of the problems discussed in the course (1 lecture)

- a. Problems in computational biology including genome assembly in different flavours, RNA-seq, Chip-seq, ATAC-seq, 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. Sequencing technologies (short read technologies like Illumina, long read technologies like Pacific Biosciences and Oxford Nanopore, linked read technologies like 10x)
- b. Guest lecture by <u>Stephen Turner</u>, Co-founder and Chief Technology Officer, <u>Pacific Biosciences</u> on 13 April 2016
- 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)
Use	eful Resources
1.	Ben Langmead's lecture notes
2.	Bioinformatics algorithms by Compeau and Pevzner