

## HGEN 663: Beyond the Human Genome

### Lectures

January 9 – April 10, 2019

Wednesdays 9:05 – 11:55

Genome 7213

### Instructor

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Genome 7210

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### TA

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### Overview

HGEN663 will be an analysis-focused course, with an emphasis on using bioinformatics tools through basic command line inputs on Unix servers and the R programming language. Because of the computer-based nature of the course, everyone will be **required to have a laptop computer**.

Most sessions will begin with an introductory lecture, followed by some practical presentations by the instructor and TA, and finish with simple hands-on exercises to be done by the students. There will be small take-home assignments to be done by the students to ensure everyone is keeping up, and also 3 larger assignments that will require an analysis of a “realistic” dataset, drawing conclusions, and reporting on final result.

### Course evaluation

- ▶ **Short take home assignments 30% (approximately 3% each assignment)**
- ▶ **Long assignments 60% (20% per assignment)**
- ▶ **Class participation 10%:**

This grade represents student contribution to class participation during open discussion and hands on exercises in class. There will be some assigned readings, mainly current research papers illustrating the biological issues and genomic techniques, and students are expected to have critically read the assigned material, be prepared to ask questions aiming to clarify the strengths and weaknesses of the approaches. Since this is an attendance-based course, attendance is required. A student is allowed to miss one class with no penalty. Additional absences, even if motivated will reduce the participation grade. A student cannot miss more than 3 classes.

**Tentative schedule**

- ▶ **January 9<sup>th</sup>: Introduction to Genomics**
  - ▷ Sequencing the Human Genome
  - ▷ High throughput DNA sequencing
  - ▷ Basic sequence data processing, alignment
  - ▷ Accessing resources, R
- ▶ **January 16<sup>th</sup>: Human Genetic Variation**
  - ▷ Variant calling
  - ▷ Quality control and evaluation
  - ▷ Visualization
- ▶ **January 23<sup>rd</sup>: Exome sequencing in Human Disease**
  - ▷ Variant annotation
  - ▷ Functional interpretation
  - ▷ Filtering
- ▶ **January 30<sup>th</sup>: Whole Genome Sequencing and analysis**
  - ▷ Copy number variant detection
  - ▷ Structural variation
  - ▷ Non-coding DNA
- ▶ **February 6<sup>th</sup>: Gene expression analysis**
  - ▷ Transcriptome
  - ▷ RNA sequencing
- ▶ **February 13<sup>th</sup>: Comparative analysis of gene expression data**
  - ▷ Identifying differentially expressed genes across samples
  - ▷ Visualization of gene expression results in genomics viewer
- ▶ **February 20<sup>th</sup>: More Advanced RNA-seq analysis**
  - ▷ Alternative splicing
  - ▷ Detection of novel transcripts, non-coding RNAs
  - ▷ Identification of fused transcripts in cancer
  - ▷ Variant calling
- ▶ **February 27<sup>th</sup>: Transcriptome analysis**
  - ▷ Principal component analysis, clustering, classification
  - ▷ Pathway and geneset enrichment analysis
- ▶ **March 6<sup>th</sup>: No class**
- ▶ **March 13<sup>th</sup>: Epigenetics, DNA methylation and analysis**
  - ▷ Identification of differentially methylated bases
  - ▷ Differentially methylated regions
- ▶ **March 20<sup>th</sup>: Epigenetics, chromatin remodeling, histone modifications**
  - ▷ Analysis of ChIP-seq data

- ▷ Peak calling
- ▶ **March 27<sup>th</sup>: Analysis of epigenomic data**
  - ▷ Identifying features such as promoters, enhancers
  - ▷ Identifying differentially used promoters
  - ▷ Regulatory motifs
- ▶ **April 3<sup>rd</sup>: Integrative genomics**
  - ▷ Statistical analysis of various genomic data types
  - ▷ Visualization of results
- ▶ **April 10<sup>th</sup>: ?**
  - ▷ ?

**Academic integrity**

McGill University values academic integrity. Therefore, all students must understand the meaning and consequences of cheating, plagiarism and other academic offenses under the Code of Student Conduct and Disciplinary Procedures. Additional details can be found [here](#).

**Language policy**

In accord with McGill University's Charter of Students' Rights, students in this course have the right to submit in English or in French any written work that is to be graded. Additional details can be found [here](#).