

## HGEN 663: Beyond the Human Genome

### Lectures

January 4 – April 5, 2023  
Wednesdays 9:05 – 11:55  
In person, location TBD

### Instructor

Prof. Jacek Majewski Genome  
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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 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 4<sup>th</sup>: Introduction to Genomics**
  - Sequencing the Human Genome
  - High throughput DNA sequencing
  - Basic sequence data processing, alignment
  - Accessing resources, R
- **January 11<sup>th</sup>: Human Genetic Variation**
  - Variant calling
  - Quality control and evaluation
  - Visualization
  - Variant annotation
  - Functional interpretation
- **January 18<sup>th</sup>: Whole Genome Sequencing and analysis**
  - Copy number variant detection
  - Structural variation
  - Non-coding DNA
- **January 25<sup>rd</sup>: Long Read and Single Molecule Sequencing**
  - TBD
- **February 1<sup>st</sup>: Transcription and RNA Sequencing**
  - Transcriptome
  - RNA-sequencing
- **February 8<sup>th</sup>: Transcriptome Analysis**
  - Identifying differentially expressed genes across samples
  - Visualization of gene expression results in genomics viewer
- **February 15<sup>th</sup>: More Advanced RNA-seq analysis**
  - Alternative splicing
  - Detection of novel transcripts, non-coding RNAs
  - Identification of fused transcripts in cancer B Variant calling
- **February 22<sup>nd</sup>: Single Cell Analysis**
  - Guest Speaker Dr. Gael Cagnone

- **March 8<sup>th</sup>: Epigenetics, DNA methylation and analysis (note, this is during the McGill reading week)**
  - Identification of differentially methylated bases
  - Differentially methylated regions
- **March 15<sup>th</sup>: Epigenetics, chromatin remodeling, histone modifications**
  - Analysis of ChIP-seq data
  - Peak calling
- **March 22<sup>nd</sup>: Analysis of epigenomic data**
  - Identifying features such as promoters, enhancers
  - Identifying differentially used promoters
  - Regulatory motifs
- **March 29<sup>th</sup>: Chromatin Conformation, HiC**
  - Guest Speaker Dr. Josee Dostie
- **April 5<sup>th</sup>: Integrative Analysis, Retrospective, Overview**
  - TBD

**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](#).