Course Code: **MMG1305H S**  
**Course Title:** Comparative and Population Genomics - from Model Organisms to Humans  
**Course Coordinator:** Lincoln Stein, Zhaolei Zhang  
**Course Location:** TBD  
**Course Times:** Wednesdays 1-3pm  
**Course Dates:** 03/27, 04/03, 04/10, 04/17, 04/24, 05/01

### Overview

Comparative genomics is the analysis and comparison of genes and genomes from different species. Comparisons among evolutionarily related species can often help to identify functionally important and evolutionarily conserved genes or regulatory elements. In addition to protein and DNA sequences, comparative analysis can be also extended to gene expression profiles, protein-protein interactions, genetic interactions, and regulatory interactions, which in turn help us understand the origin, evolution, and importance of these important cellular interactions.

In contrast to inter-species comparisons, population genomics compares the gene and genome sequences, gene expression or regulation among individuals within a population of the same species. For example, population genomics allows us to determine the level of genetic heterogeneity (variation) in the human population, and how such genetic variation correlates with both gross and molecular phenotypes.

In this course, we will introduce the concepts, computational and experimental methods, databases, and tools that are used in comparative and population genomics. The course will be a mixture of lectures and student presentations, where classical and contemporary papers will be discussed.

### Course Objectives:

- Introduce fundamental concepts and techniques in molecular evolution and population genetics.
- Explore computational models to detect regions in the genome that are under positive or negative selection.
- Understand human genetic variation.
- Understand GWAS and related techniques
- Evolution in regulatory elements and gene expression
- Understand cancer as an evolutionary process

### Marking Scheme:

- 15% for class participation  
- 25% for paper presentation  
- 60% for a final project – a five-page CIHR grant LOI

If you anticipate missing a class you must let the instructor know in advance, given the weight on participation and the fact that there are only six classes. Providing that you had a legitimate reason for missing the class, you will be provided with an assignment based on the reading for that week that you can use to make up for the lost class.
The basic outline for what will be covered in the six weeks is below. Assigned reading will be sent out the week in advance. In addition to the research articles, a review article may be distributed that is meant to provide a bit of context for the lecture for those students with less background and will not be a specific point of discussion.

Week 1: Course Introduction, Molecular Evolution (March 27, 2024)

Week 2: Genome sequence conservation: PhastCons and other tools (April 03, 2024)

Week 3: Comparing genomes: finding disease causing variants (April 10, 2024)

Week 4: Tumor mutation signatures and clonal evolution (April 17, 2024)

Week 5: Tumor driver mutations (April 24, 2024)

Week 6: Evolution in gene expression and gene regulation (May 01, 2024)