

Molecular Genetics Graduate Topic Course

Course Title: Human Genome Analysis

Course Location: Peter Gilgan Centre for Research & Learning (PGCRL), room TBD

Course Time and Date: Thursdays 2pm-4pm (March 20, 2025 – April 24, 2025 inclusive)

Course Instructor(s): Ryan Yuen and Gregory Costain

Instructor Contact Information (email): ryan.yuen@sickkids.ca; gregory.costain@sickkids.ca

Course Overview:

The course will start with an introductory lecture, covering the basic theoretical and practical aspects of human genome analysis, including a historical perspective of advances in the field. Subsequent sessions will involve class presentations of papers covering topics related to human genome analysis. All student papers will be chosen by the coordinators to reflect the following areas.

- Genome Sequencing and Analysis
- Human Genome Variation
- Common and Rare DNA Variant Interpretation
- Other 'Omic' Approaches That Facilitate Genome Analysis

Course Objectives:

- To understand the breadth of current human genome analysis approaches
- To understand the relationship between advances in technology and advances in human genome research
- To be able to critically assess research papers that employ human genome analysis techniques

Marking Scheme:

Students will be evaluated on presentation of research papers, participation in class discussions, and on a short (2 page) News and Views type article. Grades will be determined as follows:

- In-class research paper presentation 40%
- In-class discussion 20%
- News & Views article (as evaluated by classmates) 40%

If you anticipate missing a class, you should let the instructors know in advance.

The basic outline for what will be covered in the six weeks is below. Assigned journal club papers will be sent out in advance of the course start date.

Week 1: Overview of Human Genome Analysis (March 20, 2025)

Week 2: Genome Sequencing and Analysis (March 27, 2025)

Week 3: Human Genome Variation (April 3, 2025)

Week 4: Rare DNA Variants in Health and Disease (April 10, 2025)

Week 5: Common DNA Variants in Health and Disease (April 17, 2025)

Week 6: Multi-Omic Approaches to Facilitate Genome Analysis (April 24, 2025)