**MSC2010Y “Molecular Medicine in Human Genetic Disease”**

**Course Director: Lucy Osborne** [**lucy.osborne@utoronto.ca**](mailto:lucy.osborne@utoronto.ca)

***Course Description:***This course should encourage students to develop an approach to the genetic analysis, investigation and treatment of human disease. It is a required course for first year students in the Master of Science in Genetic Counseling.

In the body of the course, a series of ~16 lectures will cover background relevant to the study of human genetic disease as well as specific examples of human genetic disease research.

Introduction to genetic and molecular approaches: Students will be introduced to useful tools and concepts that enable the study of a wide spectrum of human genetic disease. These include such topics as: quantitative genetics; genome analysis; copy number variation and the use of animal models.

Specific examples of human genetic disease research: Lecturers will be encouraged to discuss the basic clinical presentation and pathophysiology of the genetic disease that they study; the approach they take in their research; the molecular changes that occur and key findings. Each lecture will provide an example of the application of molecular tools to the investigation of a specific human disease. Lecturers will be encouraged to provide weekly reading assignments in advance of the lecture and will usually provide their slides to the class for review.

Each student will be required to sit a short answer exam (December), prepare a Letter of Intent for a grant proposal that addresses a research topic in human genetic disease (February), give a 10-15 minute in-class slide presentation of a research paper assigned by the Course Director (March), and write a short News & Views type article about a paper or papers in the field of human genetic disease that has been published during the past calendar year (April).

**MSC2010Y Marking Scheme**

**Short Answer Examination** **20% of marks**

This will consist of answering five out of eight or nine questions related to guest lectures between September and December. The exam is 1 hour long, so the answers are short. This is not a big deal! It is just to make sure that everyone is on the same page and understands some of the important concepts of what the course is teaching. The questions and answers are provided by the lecturers themselves.

**Letter of Intent** **20% of marks**

The LOI should be on a topic which is NOT similar to a student’s current thesis research project. The aim of this assignment is to make students think about things other than their thesis work, so they should try to choose an area that is outside their comfort zone.

Students are strongly encouraged to talk to a PI or other colleagues about their ideas. They are free to contact any of the lecturers on the course who will be happy to discuss the proposal and give advice on specifics that they are familiar with.

This is a 1-page assignment that usually precedes the submission of a full grant proposal.

The LOI should be a maximum of 1 single spaced page. It should include some background to the project, a clearly stated hypothesis, brief account of how the hypothesis will be addressed, a summary of expected findings and its significance to the research community.

The object of the LOI is to give the granting agency (in this case the course director) a tantalizing glimpse of the proposed research and help them assign reviewers. For some funding bodies an LOI is used to select those projects that go forward to full applications. References should NOT be included.

The title and content of the LOI will be discussed in class two weeks before the submission deadline to help students round out their ideas and refine their aims and methods.

**In-Class Journal Club** **20% of marks**

Over the last five sessions of the course, each student will present a topical paper in a 15 minute PowerPoint slide presentation to the class, followed by discussion with the students and course director. Students should summarize the research carried out in the paper and point out the good things and perhaps bad things. A perspective on where the paper leaves us and what research should be carried out in the future would also be helpful (has anything pertinent happened since?). The papers will be chosen and assigned by the course director at the start of classes in January.

**News and Views Article**  **40% of marks**

This is a 2-page (maximum) News & Views type report that summarizes the importance of one or two papers on a topic of interest that were published in the past year.

There is a limit of 2 pages, INCLUDING references and figures. For examples look at New & Views articles in journals such as Nature or Nature Genetics. Figures are encouraged where appropriate and can be “borrowed” from other publications. References can be added in short form (eg. Nature style) to save space.

The paper(s) should be interesting to you and to a wider audience in the fields of molecular medicine and human genetics. The papers do not have to be in the super-high impact journals – there are a lot of fantastic papers in other journals too – but obviously the research must be of significant impact in the field.

The N&V article should be designed to highlight the advances made in the research and the impact to the field, as well as any caveats and future work that will build on the findings. The N&V can be formatted in any style.

MSC 2010Y Molecular Medicine in Human Genetic Disease Lecture Schedule 2024-25

Wednesdays from 3-5 pm

**Fall term: Student Commons SU 440 (**230 College Street)

**Winter term: Student Commons SU 440 (**230 College Street)

## Date Subject Lecturer

Sept 11 Course overview and check-in Lucy Osborne

Sept 18 Structural Variation of the Human Genome Christian Marshall

Sept 25 Quantitative Genetics Andrew Paterson

Oct 2 Genetic Counseling Susan Armel and Laura Redondo

Oct 9 Gene Regulation Michael Wilson

Oct 16 Schizophrenia and Polygenic Risk Jacob Vorstman

Oct 23 7q11.23 Genomic Disorders Lucy Osborne

Oct 30 Genetic Imprinting Sanaa Choufani

Nov 6 ASHG meeting – no class

Nov 13 Epigenetics in Genetic Disease James Eubanks

Nov 20 Pharmacogenetics Daniel Mueller

Nov 27 CRISPR Gene Editing Zhenya Ivakine

Dec 4 **Exam**

**HOLIDAY BREAK**

Jan 8 Cancer Cytogenetics Afia Hasnain

Jan15 Breast and Ovarian Cancers Mohammad Akbari

Jan 22 In class discussion of LOI Lucy Osborne

**Title of LOI due**

Jan 29 Cancer and Hypermutation Uri Tabori

Feb 5 Infectious Diseases Ian Crandall

Feb 12 Retinal Diseases and Therapies Brian Ballios

Feb 19 Reading week – no class

**LOI due**

Feb 26 **In-class Journal Club** Lucy Osborne

(Feb 28 **Drop Date)**

Mar 5 **In-class Journal Club** Lucy Osborne

Mar 12 **In-class Journal Club** Lucy Osborne

Mar 19 **In-class Journal Club** Lucy Osborne

Mar 26 **In-class Journal Club** Lucy Osborne

Apr 2

Apr 9 **Final Assignment due**