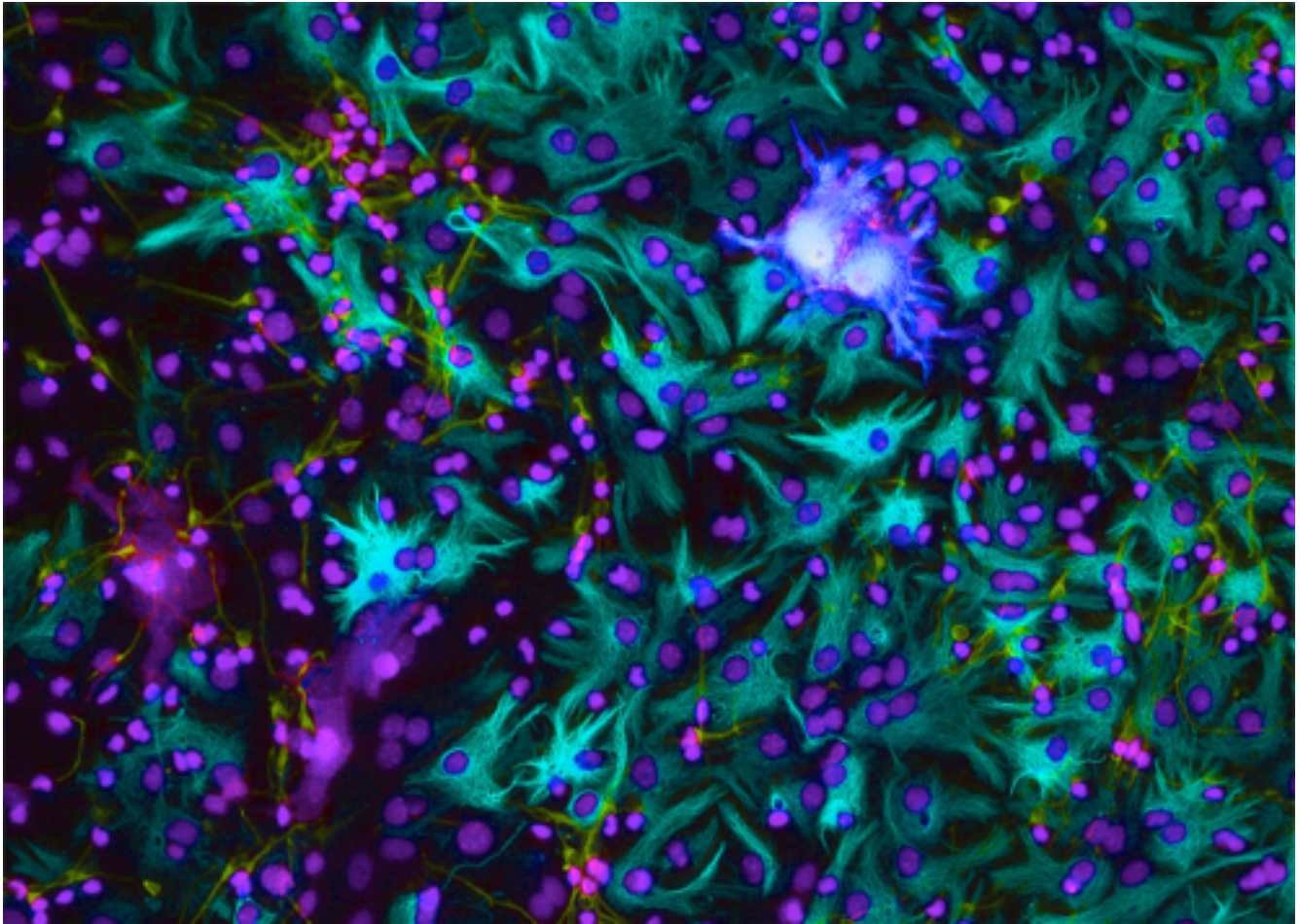

MGY250H1

An Introduction to Medical Genetics

2016 COURSEBOOK



*Neurons and astrocytes differentiated from mouse neural stem cells
photo courtesy of Samantha Yammine*



Molecular Genetics
UNIVERSITY OF TORONTO

Welcome to the inaugural session of MGY250!

We are thrilled to have you join us!

In 2016, it is difficult to read the news and not encounter a story or issue related to medical genetics. As the costs and technological limitations of genome sequencing decrease and our understanding of the genetic basis of human disease increases, we find ourselves entering an era that will revolutionize the way we think about our bodies and how we treat genetic disease. For the first time in history, we are beginning to be able to see disease looming in the distance and may be capable of halting it before it strikes. This is a fascinating time to be studying genetics!

Inherent to these big ideas are ethical considerations. As more people are impacted by medical genetics, there will be a greater need for all citizens to have a basic understanding of medical genetics.

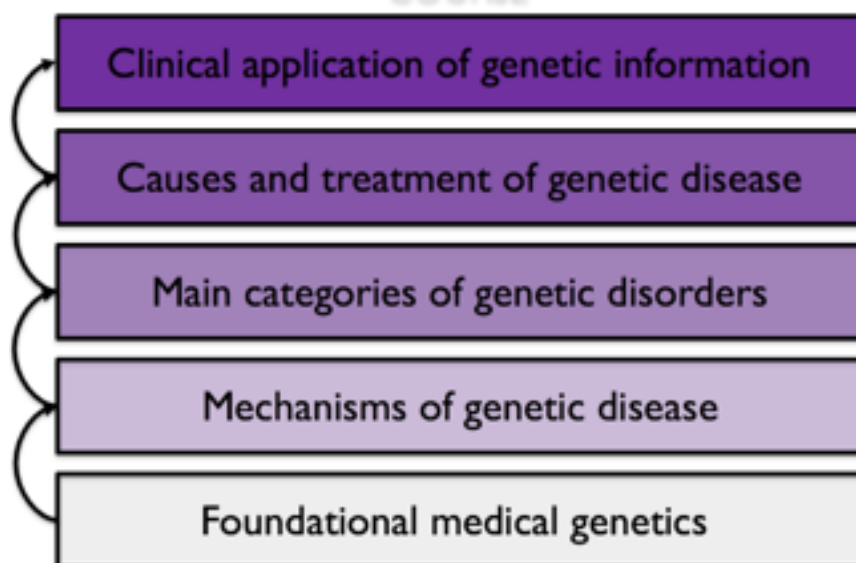
In this course we aim to provide you with a solid foundation in medical genetics, from which to build a knowledge of the mechanisms of genetic disease. We will examine the main categories of genetic disease, chromosomal, single and complex disorders. You will learn about how scientists determine the cause of genetic disease from population studies and animal models, and how these diseases are being treated. From there we will shift to look at how this information we have acquired is being implemented in the clinic. Finally we will end with a case study that takes us on the journey beginning with the initial discovery of a gene critical for the development of a childhood cancer and ending with how that information can be useful for genetic testing in Canada and abroad.

This is the first time this course is being offered and it is sure to be an adventure. Without doubt, we will hit some bumps along the way but we will do our best to be clear about our expectations and receptive to feedback. As we move along in the course, we (and the course!) will adapt to maximize what is working and eliminate what is not.

Looking forward to an exciting semester!

Sincerely,

Jessica Hill



PREREQUISITES

To succeed in MGY250, you will need a basic understanding of the cell (What is DNA? What are genes, chromosomes, proteins?) and some fundamental cellular processes (What is transcription, translation, mitosis, meiosis?).

COURSE ORGANIZATION

MGY250 is organized into “units”, each of which consist of roughly one week’s worth of material. Note that some units may be longer than others and require more time.

Each unit is composed of three main parts:

1. Videos,
2. Readings, and a
3. Unit Exam.

The videos are the core of each unit. Videos are mostly lectures from one of the course contributors.

The readings may be assigned from the textbook or supplementary material.

Some weeks, there will be an assignment due. There are a total of three assignments in this course. The assignments are designed to be progressive: for each assignment, you will be asked the same (or a very similar) question. As we progress through the course and your knowledge base grows, so should the depth of your answer.

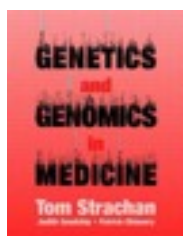
PROGRESSING THROUGH THE COURSE

In an online course, there is no scheduled lecture time. Over the course of each week, you are expected to watch the videos and do the readings assigned. However, there are deadlines only for the Unit Exams, Assignments and the Final Exam. We suggest that, like with a live course, you make sure to schedule the time in your calendar to watch the videos and do the readings assigned! Before we go further, check out this link: [Is Online Learning Right for Me?](#)

This course is wholly administered through the University of Toronto Learning Portal system, Blackboard. Logging into Blackboard will take you to the course homepage, called “Course Schedule”, where you will find the Unit Schedule and the due dates for Unit Exams and Assignments.

Units can be found in the “Units” link in the menu on the left hand side of the homepage. Assignments can be found at the “Assignments” link. Another important link is to the “Discussion Board”, where all course material related questions should be directed, and will be responded to.

As we progress through the course the units will be made available. Each unit begins with the expectations for the unit by listing what videos you are to watch and what readings are assigned. The units will also contain pdf files of all the slides in the unit video lectures. Every unit ends with a Unit Exam, typically consisting of multiple choice-type of questions. These are open book/open video exams that you can save and return to, prior to the due date.



COURSE TEXTBOOK

The textbook for this course is “Genetics and Genomics in Medicine” eds. Strachan, Goodship and Chinnery. It is available through the University of Toronto Bookstore.

The textbook is **mandatory** for the course, although the majority of your Unit Exam and Final Exam questions will come from course videos.

COURSE MARKING SCHEME

Final Exam - 40%

This is a closed book exam that will be conducted on campus (see “Final Exam” below).

Unit Exams - 25%

These are weekly quizzes focussed on the material covered in the Unit specified. The questions are designed to evaluate your background knowledge and to help the course team identify any areas of conceptual misunderstanding.

Assignments - 35%

The course assignments are progressive and designed for you to apply the knowledge that you gain in this course by arguing a position on a controversial topic

Assignment 1 - 5%

Assignment 2 - 10%

Assignment 3 - 20%

UNIT SCHEDULE

UNIT	RELEASE DATE	TOPIC	INSTRUCTORS	TA
1	Sep 12	Perspectives on medical genetics	Jessica Hill	Jessica Hill
2	Sep 19	Human genome structure and function	Michael Wilson	Armande Ang Houle
3	Sep 26	Human genome variation	James Dowling Christopher Pearson	Lindsay Smith
4	Oct 3	Pathogenic phenotypes - single gene disorders	James Dowling	Lindsay Smith
5	Oct 11	Pathogenic phenotypes - complex disease	Johanna Rommens	Armande Ang Houle
6	Oct 17	Population analysis in complex disease	Fritz Roth Philip Awadalla	Armande Ang Houle

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7	Oct 24	Using model organisms to understand disease	Jessica Hill Sean Egan	Lindsay Smith
8	Oct 31	Treating genetic disease	Derek van der Kooy Ronald Cohn	Emily Beckett Sward
9	Nov 9	Genetic testing	Maian Roifman David Chitayat George Charames Michael Szego	Emily Beckett Sward
10	Nov 14	Genetic counselling	Cheryl Shuman Andrea Shugar Randi Zlotnik Shaul	Emily Beckett Sward
11	Nov 21	Case study: childhood cancer genetics	Brenda Gallie Kahaki Kimani Helen Dimaras	Jessica Hill

IMPORTANT DATES IN THE COURSE

- Units are released on a weekly basis
- Each Unit has a Unit Exam that is released at the end of the week
- Unit Exams are due a week after they are released
- There are three assignments, each with three sections due periodically over the course

Date	DESCRIPTION
Sep 12	Unit 1 released
Sep 16	Unit 1 Exam released
Sep 19	Unit 2 released
Sep 23	Unit 2 Exam released
Sep 23	Unit 1 Exam due at 5pm
Sep 26	Unit 3 released
Sep 29	Assignment 1a — Rough draft due at 11:59pm
Sep 30	Unit 3 Exam released
Sep 30	Unit 2 Exam due at 5pm
Oct 3	Assignment 1b — Peer review due at 11:59pm
Oct 3	Unit 4 released
Oct 6	Assignment 1c — Final Report due at 11:59pm

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Oct 7	Unit 4 Exam released
Oct 7	Unit 3 Exam due at 5pm
Oct 10	Unit 5 released
Oct 14	Unit 5 Exam released
Oct 14	Unit 4 Exam due at 5pm
Oct 17	Unit 6 released
Oct 21	Unit 6 Exam released
Oct 21	Unit 5 Exam due at 5pm
Oct 24	Assignment 2a — Rough draft due at 11:59pm
Oct 24	Unit 7 released
Oct 28	Assignment 2b — Peer review due at 11:59pm
Oct 28	Unit 6 Exam due at 5pm
Oct 28	Unit 7 Exam released
Oct 31	Unit 8 released
Nov 1	Assignment 2c — Final Report due at 11:59pm
Nov 4	Unit 8 Exam released
Nov 4	Unit 7 Exam due at 5pm
Nov 7	Unit 9 released
Nov 11	Unit 9 Exam released
Nov 11	Unit 8 Exam due at 5pm
Nov 14	Unit 10 released
Nov 18	Unit 10 Exam released
Nov 18	Unit 9 Exam due at 5pm
Nov 21	Unit 11 released
Nov 25	Assignment 3a — Rough draft due at 11:59pm
Nov 25	Unit 11 Exam released
Nov 25	Unit 10 Exam due at 5pm
Nov 30	Assignment 3b — Peer review due at 11:59pm
Dec 2	Unit 11 Exam due at 5pm

Dec 6

Assignment 3c — Final Report **due at 11:59pm**

HOW TO GET HELP

Have a question? Chances are you're not the only one.

FOR QUESTIONS REGARDING COURSE MATERIAL:

We would like to have open lines of communication, so we will answer questions related to course content on the "Discussion Board" on Blackboard. ***All course material related questions should be directed to the "Discussion Board", not to your TA or the course coordinator's email.***

The "Discussion Board" is meant to be a positive space where students should feel comfortable asking questions that they have taken the time to consider independently, and for other students to provide feedback. Each forum will be monitored by a TA and/or the course coordinator, who can helpfully and respectfully clear up any misconceptions that may arise.

Each unit of the course will have its own forum. Before posting a question, please read the other posts in that forum because your question may have already been asked.

For the purposes of marking, what we say on the "Discussion Board" is the final word. If we have given incorrect information we will make an announcement to the entire class at once about where the issue was and how we will resolve it.

FOR QUESTIONS OR HELP ON OTHER ISSUES:

(eg. sick, family issues, prerequisites, etc.)

Please contact your designated contact below. You can access their contact information through the "Contacts" link on the sidebar in Blackboard:

Last name beginning with:	Contact the following:
A - Do	Jessica Hill
Du - Leong	Armande Ang-Houle
Li - Ruste	Emily Beckett-Sward
Sa - Z	Lindsay Smith

Your TA or the course coordinator will try to respond to you as quickly as possible to let you know your message has been received. However unless the need is truly urgent your matter will not be discussed and a decision will not be reached until our weekly TA meeting.

COURSE POLICIES

LATE SUBMISSIONS:

Late submissions of Unit Exams and Assignments receive a mark of 0. It is especially critical that your assignments are submitted on time in order to receive peer feedback.

SUBMISSION METHODS:

Assignments will be submitted through peerScholar only. There is also a survey requirement for the assignments, administered through Blackboard.

TECHNOLOGY REQUIREMENTS:

You must have access to a computer or a tablet with a Wi-Fi internet connection (or faster) to be able to watch the videos.

This course requires the use of computers, and of course sometimes things can go wrong when using them. You are responsible for ensuring that you maintain regular backup copies of your files, use antivirus software (if using your own computer), and schedule enough time when completing an assignment to allow for delays due to technical difficulties. Computer viruses, crashed hard drives, broken printers, lost or corrupted files, incompatible file formats, and similar mishaps are common issues when using technology, and are not acceptable grounds for a deadline extension.

EMAIL COMMUNICATION:

At times, we may decide to send out important course information by email. To that end, all students are required to have a valid University of Toronto email address. You are responsible for ensuring that your University of Toronto email address is set up and properly entered in the ROSI system.

INSTITUTIONAL POLICIES AND SUPPORT

POLICY ON DISTRIBUTION OF THE MATERIALS OUTSIDE OF THE COURSE:

At the University of Toronto and the Department of Molecular Genetics we take pride in the fact that we have unique, high-level and up-to-date expertise in the course topics. All course materials are the Intellectual Property of the lecturers. Further distribution of the lecture materials without permission constitutes an academic offence, and the instructors have the right to pursue disciplinary action.

ACADEMIC INTEGRITY:

Academic integrity is essential to the pursuit of learning and scholarship in a university, and to ensuring that a degree from the University of Toronto is a strong signal of each student's individual academic achievement. As a result, the University treats cases of cheating and plagiarism very seriously. The University of Toronto's Code of Behaviour on Academic Matters (www.governingcouncil.utoronto.ca/policies/behaveac.htm) outlines the behaviours that constitute academic dishonesty and the processes for addressing academic offences. Potential offences include, but are not limited to:

In papers and assignments:

1. Using someone else's ideas or words without appropriate acknowledgement.
2. Submitting your own work in more than one course without the permission of the instructor.
3. Making up sources or facts.
4. Obtaining or providing unauthorized assistance on any assignment.

On tests and exams:

1. Using or possessing unauthorized aids.
2. Looking at someone else's answers during an exam or test.
3. Misrepresenting your identity.

In academic work:

1. Falsifying institutional documents or grades.
2. Falsifying or altering any documentation required by the University.

All suspected cases of academic dishonesty will be investigated following procedures outlined in the Code of Behaviour on Academic Matters. If you have questions or concerns about what constitutes appropriate academic behaviour or appropriate research and citation methods, you are expected to seek out additional information on academic integrity from your instructor or from other institutional resources (see www.utoronto.ca/academicintegrity/resourcesforstudents.html).

FINAL EXAM:

The University of Toronto, St. George, Arts and Science final examination will require your attendance on Campus.

If you are eligible and require off-site proctoring, please notify your Faculty registrar and submit your request no later than twelve (12) business days after the start of term.

If requested on time, the Arts and Science Registrar will endeavour to provide arrangements for proctored exam writing for students residing more than 125 km travel distance from the campus at a proposed outside examination centre. You must provide the contact information of an institution in your area offering proctoring services, however, please note that the requested location is not guaranteed and an alternative test centre may be identified. Students are responsible for any fees charged by the test centre. Please contact the Faculty Registrar's Office for further details.

For more information see [FAQs for Off-Site Exams](#).

ACCESSIBILITY NEEDS:

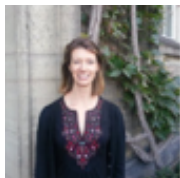
The University of Toronto is committed to accessibility. If you require accommodations for a disability, or have any accessibility concerns about the course, the classroom or course materials, please contact [Accessibility Services](#) as soon as possible.

SERVICES AND SUPPORT:

The following are some important links to help you with academic and/or technical service and support

- General student services and resources at [Student Life](#)
- Full library service through [University of Toronto Libraries](#)
- Resources on conducting online research through [University Libraries Research](#)
- Resources on academic support from the [Academic Success Centre](#)
- Learner support at the [Writing Centre](#)
- Information about [Accessibility Services](#)
- Information for [Technical Support/Blackboard Support](#) (Portal Info)

COURSE COORDINATOR AND TEACHING ASSISTANTS

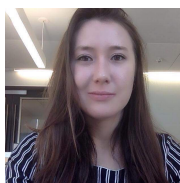


Jessica Hill

Course Co-ordinator

jessica.hill@mail.utoronto.ca

Dr. Hill completed her undergraduate studies at Queen's University and her MSc at the University of British Columbia with Sally Otto. She received her PhD from the University of Toronto in 2014, supervised by Leah Cowen. Dr. Hill then pursued post-doctoral research with Helen Dimaras at SickKids, studying challenges in implementing clinical genetics services in Canada and Kenya. Besides science, Jessica is passionate about yoga, ultimate and baking.



Armande Ang Houle

Teaching Assistant

armande.anghoule@mail.utoronto.ca

Before joining the Awadalla lab at the University of Toronto as a PhD student in 2015, Armande was awarded a BSc and a MSc in bioinformatics from the University of Montreal. In the context of her MSc, Armande created methods to analyze the allelic variation of polymorphic repetitive genes from Next-Generation Sequencing data. Armande is broadly interested in the integration of heterogeneous sources of biological data. The goal of her dissertation is to further understand the expression of meiosis specific genes, especially those involved in recombination processes. Armande aims to investigate how these genes influence the genomic landscape across different cancer types. She also likes to run long distances slowly and to chill with her two cats.

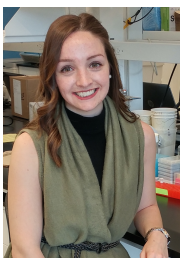


Emily Beckett Sward

Teaching Assistant

emily.beckett@mail.utoronto.ca

Emily is in her 5th year of the PhD program in Molecular Genetics. As a member of the Culotti lab, she uses the genetics of *C. elegans* to study the role of the netrin pathway in cell migration and axon guidance.



Lindsay Smith

Teaching Assistant

lin.smith@mail.utoronto.ca

Lindsay is a Master's student in the department of Molecular Genetics. Having a strong interest in muscle development and function, she is interested in the mechanisms that lead to muscle weakness in patients with various skeletal muscle diseases. Under the supervision of Dr. James Dowling, she aims to understand the role of membrane lipids, called phosphoinositides, in a component of the muscle important for excitation-contraction coupling. When not in the lab, you're likely to find her in a hockey arena or at a baseball diamond.

COURSE CONTRIBUTORS



[Philip Awadalla](#)



[George Charames](#)



[Ronald Cohn](#)



[David Chitayat](#)



[Helen Dimaras](#)



[James Dowling](#)



[Sean Egan](#)



[Brenda Gallie](#)



[Kahaki Kimani](#)



[Christopher Pearson](#)



[Maian Roifman](#)



[Johanna Rommens](#)



[Fritz Roth](#)



[Cheryl Shuman](#)



[Andrea Shugar](#)



[Michael Szego](#)



[Derek van der Kooy](#)



[Michael Wilson](#)



[Randi Zlotnik Shaul](#)