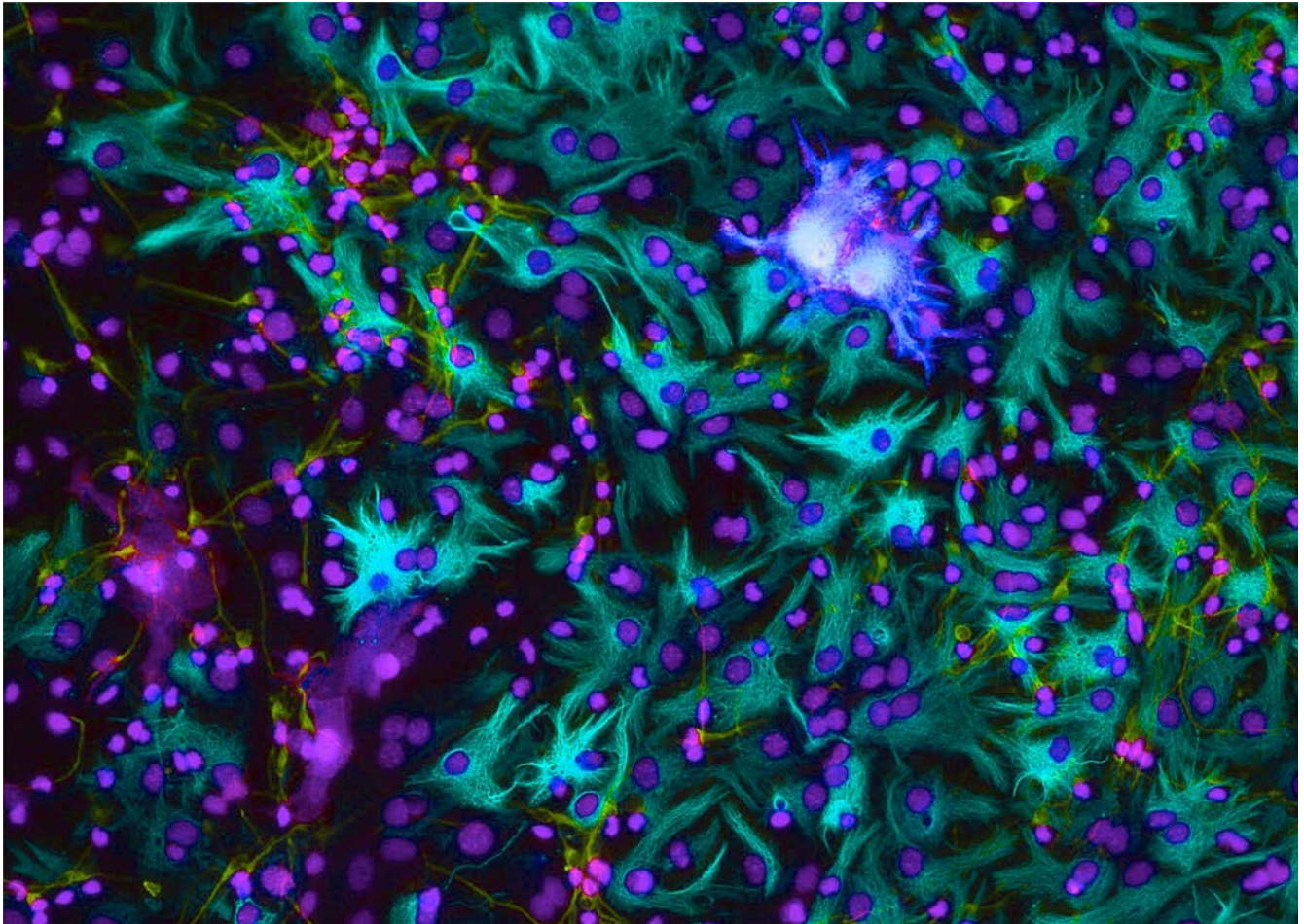

MGY250H1

Introduction to Medical Genetics

WINTER 2022 COURSEBOOK



Neurons and astrocytes differentiated from mouse neural stem cells

photo courtesy of Samantha Yammine



Molecular Genetics
UNIVERSITY OF TORONTO

Welcome to MGY250!

We are thrilled to have you join us!

In this day and age, 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.

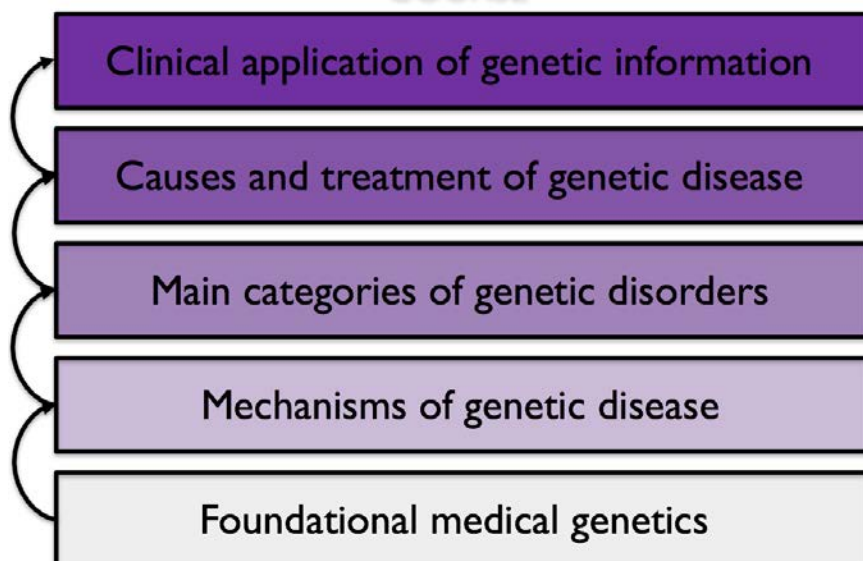
This course is sure to be an adventure. Without doubt, we will hit some bumps along the way but we will do our best to be clear about our expectations and receptive to feedback. As we move along in the course, we (and the course!) will adapt to maximize what is working and eliminate what is not.

Looking forward to an exciting semester!

Sincerely,

Jessica Hill

Assistant Professor, Teaching Stream



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 MARKING SCHEME

Final Exam — 35%

Unit Quizzes — 25%

Assignments — 30%

Assignment 1 — 4%

Assignment 2 — 10/15%**

Assignment 3 — 10/15%**

Mind maps — 11%

****the greater of your marks for Assignment 2 and Assignment 3 will be worth 15%, and the lesser will be worth 10%**

COURSE COMPONENTS

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. Nearly each week, a new unit becomes available.

Each unit is composed of four main parts:

1. **Videos**
2. **Readings,**
3. a **Discussion** page, and a
4. **Unit Quiz.**

In addition, three **assignments** are released over the duration of the course. You will also be asked to generate a **mind map** for each unit of the course.

VIDEOS

The videos are the core of each unit. Videos are mostly lectures from one of the course contributors. The units will also contain pdf files of all lecture slides.

READINGS

The readings may be assigned from the textbook or supplementary material. Most of the readings are recommended, not required. Testable material comes primarily from your video lectures, but the readings may help you put those lectures into context.

DISCUSSION BOARDS

On the discussion board for the unit, you can post any questions you have about the course material (without giving away the answers to ongoing quizzes or those that haven't yet been returned). The discussion boards are meant to be positive spaces where students should feel comfortable asking questions that they have taken the time to consider independently, and for other students to provide feedback and/or learn from their peers' questions. Before posting a question, please read the other posts on that board because your question may have already been asked. All posts should be polite and respectful. Each discussion board will be monitored by a TA and/or the course coordinator, who can help to answer any questions that may arise. There is also a general discussion board for administrative/miscellaneous questions.

UNIT QUIZZES

Unit Quizzes 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. You have 1 attempt per Unit Quiz.

ASSIGNMENTS

Some weeks, there will be an assignment due. There are a total of three assignments in this course, they can be found on the Modules page. The assignments are designed to be progressive. As we progress through the course and your knowledge base grows, so should the depth of your answer. You can find the due dates for Assignments on the course calendar.

Assignments are administered through PeerScholar, which you can access through the Assignment page in Quercus. Each assignment consists of three parts: a rough draft, peer review and a final draft. Therefore, each assignment has three due dates. It is important to submit each stage of the assignment on time so that you can participate in the subsequent stage.

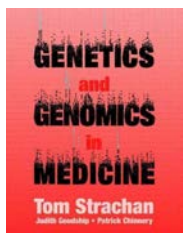
MIND MAPS

For each unit of the course, you are asked to generate a Mind Map using the tool Miro. A mind map is a visual tool to help you organize your thoughts about the lecture content for each unit and to more easily see the connections between concepts within a unit. Your mind map will be marked as a contribution mark: 1% for each unit for a total of 11% of your final mark.

COURSE CALENDAR

Important dates for this course can be found on the course calendar, such as release dates and due dates for Unit Quizzes and Assignments. Please ensure you are keeping up with due dates in the course via the course calendar.

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 **recommended** for the course, although the majority of your Unit Exam and Final Exam questions will come from course videos.

UNIT SCHEDULE

UNIT	TOPIC	INSTRUCTORS	TA
1	Perspectives on medical genetics	Jessica Hill Michael Wilson	Jessica Hill
2	Human genome structure and function	Michael Wilson	Nika Maani
3	Human genome variation	James Dowling	Vida Erfani
4	Pathogenic phenotypes - single gene disorders	James Dowling	Nika Maani
5	Pathogenic phenotypes - complex disease	Jessica Hill	Vida Erfani
6	Population analysis in complex disease	Fritz Roth Philip Awadalla	Nika Maani
7	Using model organisms to understand disease	Jessica Hill Sean Egan	Vida Erfani
8	Treating genetic disease	Derek van der Kooy Zhenya Ivakine	Nika Maani
9	Genetic testing	Maian Roifman George Charames Michael Szego	Vida Erfani
10	Genetic counselling	Cheryl Shuman Andrea Shugar Randi Zlotnik Shaul	Jessica Hill
11	Case study: childhood cancer genetics	Brenda Gallie Kahaki Kimani Helen Dimaras	Jessica Hill

IMPORTANT DATES IN THE COURSE

The release dates and due dates for all Unit Quizzes and Assignments have been added to the course calendar. Please check the calendar for the most recent updates of course due dates.

All Unit Quizzes and Assignments will be due at 11:59pm

HOW TO GET HELP

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 page for each Unit on Quercus. ***All course material-related questions should first be directed to the Unit's Discussion page.***

The Unit Discussion pages are meant to be positive spaces 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 Discussion page. 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 Unit Discussion page 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.

Last name beginning with:	Contact the following:
A — Kr	Vida Erfani vida.erfani@mail.utoronto.ca
Ku — Z	Nika Maani nika.maani@sickkids.ca

Your TA or the course coordinator will respond to you within **24 hours (excluding weekends)** 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

If you are unable to submit your Unit Quiz by the deadline and would like the opportunity to submit it up to one week following the deadline, please contact your TA.

Each course assignment has three parts — rough draft, peer review and final draft — and therefore three due dates. To participate in the review process, ***it is critical to submit your rough draft and peer review by their due dates***. Extensions will not be granted for the first two parts of each assignment, barring exceptional circumstances. If you feel you will have trouble meeting those deadlines, we can give you an extension on your Unit Quiz for that week so that you can prioritize completing your assignment. Please contact your TA if that is support you require.

SUBMISSION METHODS

Assignments will be submitted through Quercus only. Quercus will save your Unit Quiz responses as you go along, but be careful — if your Internet cuts out, you may lose answers. Your Unit Quiz will be automatically submitted at the Due Date.

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 or by announcement. 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 ACORN 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 coordinator



Dr. Jessica 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, having studied the evolution of drug resistance in the fungal pathogen *Candida albicans* with Leah Cowen. She then pursued post-doctoral research with Helen Dimaras at SickKids, studying clinical cancer genetics in global health. When not reading about science, Jessica can be found baking or on her Peloton or playing with her toddler.

Vida Erfani



Hi, I'm Vida! I did my undergraduate at U of T in molecular genetics and human physiology, took a gap year to work in a research lab, and I'm now a third-year molecular genetics PhD student in Dr. Brian Ciruna's lab! Our lab studies scoliosis, using zebrafish as a model organism. Therefore, my day-to-day life as a grad student involves studying scoliotic fish spines: reading about fish spines, thinking about fish spines, and dreaming about fish spines. When I'm not in the lab, I can be found sewing a new dress, in ballet class, or making memes of my lab mates.

Nika Maani



Nika Maani was born in Ottawa but lived in both of Canada's largest cities. After completing her undergraduate studies at McGill University in Montreal, she moved to Toronto for grad school. Here, she specialized in therapy development for rare neuromuscular diseases in children. She completed her MSc. in 2018 and is now working towards her PhD in the same field. She is a passionate communicator and loves to teach others, especially about the benefits of grad school. Outside the lab, she often finds herself immersed in home organization and DIY projects.