



MBG*3050 Human Genetics

Winter 2024

Section(s): 01

Department of Molecular and Cellular Biology

Credit Weight: 0.50

Version 1.00 - December 18, 2023

1 Course Details

1.1 Calendar Description

This course is designed to introduce the student to the study of biological inheritance in humans. The course includes discussion of the genetic basis of human individual differences, gene frequencies in human populations, human behavioral genetics, human cytogenetics, biochemical genetics and developmental genetics, medical genetics and other aspects of human heredity.

Pre-Requisites: MCB*2050

1.2 Course Description

This course is designed to provide students with advanced study into biological inheritance in humans. Topics include the genetic basis of individual differences, human origins, developmental genetics, cancer genetics and aging. We will also discuss research strategies for identifying and studying genes responsible for diverse functions and traits, as well as approaches to treating human disease, personalized medicine and the application and consequences of modern genetic technologies in our society. This course will consist of a series of lectures and presentations by student groups, as well as complementary laboratory modules.

1.3 Timetable

Lectures:

Monday, Wednesday and Friday from 11:30 AM -12:20 PM. Lecture notes will be posted on CourseLink in advance.

Presentations:

Presentations will be held virtually, via Zoom.

Laboratories:

Alternating weeks on Tuesday or Wednesday from 2:30-5:20 PM. See Courselink for details.

1.4 Final Exam

April 16th, 2024. 11:30AM-1:30PM

Room TBA

2 Instructional Support**2.1 Instructional Support Team**

Instructor: Nina Jones
Email: jonesmcb@uoguelph.ca
Telephone: +1-519-824-4120 x53643
Office: SSC 3461
Office Hours: Dr. Jones can be contacted via email to arrange.

Course Co-ordinator: Kelsey Pannunzio
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Telephone: 519-824-4120 Ext. 56349
Office: SSC 3253

2.2 Teaching Assistants

Teaching Assistant (GTA): Una Pantic
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Teaching Assistant (GTA): Alice Wang
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Teaching Assistant (GTA): Olivia Bebenek
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3 Learning Resources**3.1 Required Resources**

Courselink (Website)

<https://courselink.uoguelph.ca>

This course will make use of the University of Guelph's Courselink website. Consequently, you are responsible for all information posted on the Courselink page for MBG*3050. Please check it regularly.

Human Genetics Lab Manual (will be provided to you) (Lab Manual)

- Lab manuals will be available as free PDFs on Courselink. You are responsible for either printing the manual (recommended) or accessing it on a laptop/tablet in-lab. **You must bring the manual (either printed or digital) to each lab session.**
- These contain detailed experimental protocols in addition to background information on lab topics.

3.2 Recommended Resources

Human Molecular Genetics 4e or 5e (Textbook)

Recommended Textbook: Tom Strachan and Andrew P. Read, 2011. *Human Molecular Genetics* (4th edition*). (HMG4) Garland Publishing.

- Copies of the 4th edition are available in the UofG Bookstore and on 2-hour reserve at McLaughlin Library.
- *Please note that a 5th edition was released in December 2018 and is available for purchase through online retailers. Updates from this text will be incorporated.

3.3 Additional Resources

Reserve material (Textbook)

Additional textbooks available on 2-hour reserve at McLaughlin Library:

- Nussbaum, McInnes, Willard, 2007. *Genetics in Medicine*. Saunders Elsevier
- Scott Gilbert, 2010. *Developmental Biology* (9th edition). Sinauer Associates.
- Ian Young, 2005. *Medical Genetics*. Oxford University Press.

4 Learning Outcomes

4.1 Course Learning Outcomes

By the end of this course, you should be able to:

1. • Describe the nature of genetic material

- Understand transmission/patterns of inheritance
- Discuss the molecular biology underpinning gene function
- Describe mechanisms of gene expression and regulation
- Explain how genetic variation arises
- Describe processes of population genetics that influence evolution
- Understand the benefits and limitations of using model organisms in genetics
- Explain modern methods and tools in genetics

Students should also be able to:

- Locate, read and comprehend primary research papers on genetics
- Design experiments using appropriate controls and sample sizes
- Apply statistical methods when analysing data
- Generate and interpret graphs displaying experimental results
- Communicate experimental findings in lab reports and oral presentation
- Effectively explain genetics concepts to different audiences
- Identify and critique scientific issues relating to society or ethics

The learning outcomes for this course have been developed in line with core concepts and competencies established by the Genetics Society of America Education Committee.

5 Teaching and Learning Activities

5.1 Lectures

Lectures will be presented using PowerPoint and will be posted on Courselink prior to the lecture time.

5.2 Laboratories

Laboratories are scheduled to run in-person. **Please note that laboratory format is subject to change depending on directives received from the University and province.** More information is available on Courselink with a complete lab schedule and assignment due dates for each section.

Attendance at all five labs is mandatory. Failure to attend all 5 labs will result in grade deductions from lab reports.

5.3 Formal scientific lab report

You are required to write two lab reports for this course. The first is to cover Lab 2, integrating and analyzing anonymized data on TAS2R38 genotype. The second is to cover Labs 4 and 5, comparing p53 gene and protein levels in normal and cancer cells. The best one out of two will be considered, worth 20% total. Instructions and rubrics are provided in Courselink.

5.4 Quizzes

Each lab will have an online quiz (available via Courselink). There will be a pre-lab quiz which must be completed **before** the start of your scheduled lab (worth 5% total, best 4 of 5 quiz grades will be tallied).

5.5 Presentation

Date and topic to be selected/assigned in the first two weeks of class (worth 10% total). Students will work in groups of four and each presentation will be ~12 minutes in length, with ~3 minutes for questions. Students will be asked to prepare a summary with key terms and points, as well as questions that can be used for the final exam. Students will sign up in the first week of the semester. Detailed instructions for the presentation will be posted in a separate document on Courselink.

5.6 Tentative lecture topics

- Chromosome structure and function
- Patterns of single gene inheritance
- Multifactorial inheritance
- Human genome variation
- Human evolutionary genetics
- Identifying human disease genes
- Early human development
- Developmental genetics
- Genetics of sex determination
- Stem cells and organoids
- Cancer genetics
- Genetic testing
- Model organisms and modelling disease
- Genetic approaches to treating disease
- Personalized medicine

5.7 Tentative lecture schedule

A detailed schedule will be posted in a separate document on Courselink.

6 Assessments

6.1 Marking Schemes & Distributions

Course Evaluation and Exam Dates:

| Assessment | Weight (% final grade) | Date/Location |
|-----------------------|------------------------|--|
| Midterm Exam | 25% | During class time (11:30 AM-12:20 PM) on February 14, 2023 |
| In-class Presentation | 10% | see schedule on Courselink, during class time |
| Lab Reports | 20% | see schedule on Courselink |
| Lab Quizzes | 5% | see schedule on Courselink |
| Final Exam | 40% | April 16, 2024. 11:30AM-1:30PM |

7 Department of Molecular and Cellular Biology

Statements

7.1 Academic Advisors

If you are concerned about any aspect of your academic program:

- Make an appointment with a program counsellor in your degree program. [B.Sc. Academic Advising](#) or [Program Counsellors](#)

7.2 Academic Support

If you are struggling to succeed academically:

- Learning Commons: There are numerous academic resources offered by the Learning Commons including, Supported Learning Groups for a variety of courses, workshops related to time management, taking multiple choice exams, and general study skills. You can also set up individualized appointments with a learning specialist.
<http://www.learningcommons.uoguelph.ca/>
- Science Commons: Located in the library, the Science Commons provides support for physics, mathematic/statistics, and chemistry. Details on their hours of operations can be found at: <http://www.lib.uoguelph.ca/get-assistance/studying/chemistry-physics-help> and <http://www.lib.uoguelph.ca/get-assistance/studying/math-stats-help>

7.3 Wellness

If you are struggling with personal or health issues:

- Counselling services offers individualized appointments to help students work through personal struggles that may be impacting their academic performance.
<https://www.uoguelph.ca/counselling/>
- Student Health Services is located on campus and is available to provide medical attention. <https://www.uoguelph.ca/studenthealthservices/clinic>
- For support related to stress and anxiety, besides Health Services and Counselling Services, Kathy Somers runs training workshops and one-on-one sessions related to stress management and high performance situations.
<http://www.selfregulationskills.ca/>

7.4 Personal information

Personal information is collected under the authority of the University of Guelph Act (1964), and in accordance with Ontario's Freedom of Information and Protection of Privacy Act (FIPPA) <http://www.e-laws.gov.on.ca/index.html>. This information is used by University officials in order to carry out their authorized academic and administrative responsibilities and also to establish a relationship for alumni and development purposes.

For more information regarding the Collection, Use and Disclosure of Personal Information policies please see the Undergraduate Calendar.
(<https://www.uoguelph.ca/registrar/calendars/undergraduate/current/intro/index.shtml>)

7.5 Course Offering Information Disclaimer

Please note that course delivery format (face-to-face vs online) is subject to change up to the first-class day depending on requirements placed on the University and its employees by public health bodies, and local, provincial and federal governments. Any changes to course format prior to the first class will be posted on WebAdvisor/Student Planning as they become available.

8 University Statements

8.1 Email Communication

As per university regulations, all students are required to check their e-mail account regularly: e-mail is the official route of communication between the University and its students.

8.2 When You Cannot Meet a Course Requirement

When you find yourself unable to meet an in-course requirement because of illness or compassionate reasons please advise the course instructor (or designated person, such as a teaching assistant) in writing, with your name, id#, and e-mail contact. The grounds for Academic Consideration are detailed in the Undergraduate and Graduate Calendars.

Undergraduate Calendar - Academic Consideration and Appeals

<https://www.uoguelph.ca/registrar/calendars/undergraduate/current/c08/c08-ac.shtml>

Graduate Calendar - Grounds for Academic Consideration

<https://www.uoguelph.ca/registrar/calendars/graduate/current/genreg/index.shtml>

Associate Diploma Calendar - Academic Consideration, Appeals and Petitions

<https://www.uoguelph.ca/registrar/calendars/diploma/current/index.shtml>

8.3 Drop Date

Students will have until the last day of classes to drop courses without academic penalty. The deadline to drop two-semester courses will be the last day of classes in the second semester. This applies to all students (undergraduate, graduate and diploma) except for Doctor of Veterinary Medicine and Associate Diploma in Veterinary Technology (conventional and alternative delivery) students. The regulations and procedures for course registration are available in their respective Academic Calendars.

Undergraduate Calendar - Dropping Courses

<https://www.uoguelph.ca/registrar/calendars/undergraduate/current/c08/c08-drop.shtml>

Graduate Calendar - Registration Changes

<https://www.uoguelph.ca/registrar/calendars/graduate/current/genreg/genreg-reg-regchg.shtml>

Associate Diploma Calendar - Dropping Courses

<https://www.uoguelph.ca/registrar/calendars/diploma/current/c08/c08-drop.shtml>

8.4 Copies of Out-of-class Assignments

Keep paper and/or other reliable back-up copies of all out-of-class assignments: you may be asked to resubmit work at any time.

8.5 Accessibility

The University promotes the full participation of students who experience disabilities in their academic programs. To that end, the provision of academic accommodation is a shared responsibility between the University and the student.

When accommodations are needed, the student is required to first register with Student Accessibility Services (SAS). Documentation to substantiate the existence of a disability is required; however, interim accommodations may be possible while that process is underway.

Accommodations are available for both permanent and temporary disabilities. It should be noted that common illnesses such as a cold or the flu do not constitute a disability.

Use of the SAS Exam Centre requires students to make a booking at least 14 days in advance, and no later than November 1 (fall), March 1 (winter) or July 1 (summer). Similarly, new or changed accommodations for online quizzes, tests and exams must be approved at least a week ahead of time.

For Guelph students, information can be found on the SAS website
<https://www.uoguelph.ca/sas>

For Ridgetown students, information can be found on the Ridgetown SAS website
<https://www.ridgetownc.com/services/accessibilityservices.cfm>

8.6 Academic Integrity

The University of Guelph is committed to upholding the highest standards of academic integrity, and it is the responsibility of all members of the University community—faculty, staff, and students—to be aware of what constitutes academic misconduct and to do as much as possible to prevent academic offences from occurring. University of Guelph students have the responsibility of abiding by the University's policy on academic misconduct regardless of their location of study; faculty, staff, and students have the responsibility of supporting an environment that encourages academic integrity. Students need to remain aware that instructors have access to and the right to use electronic and other means of detection.

Please note: Whether or not a student intended to commit academic misconduct is not relevant for a finding of guilt. Hurried or careless submission of assignments does not excuse students from responsibility for verifying the academic integrity of their work before submitting it. Students who are in any doubt as to whether an action on their part could be construed as an academic offence should consult with a faculty member or faculty advisor.

Undergraduate Calendar - Academic Misconduct
<https://www.uoguelph.ca/registrar/calendars/undergraduate/current/c08/c08-amisconduct.shtml>

Graduate Calendar - Academic Misconduct
<https://www.uoguelph.ca/registrar/calendars/graduate/current/genreg/index.shtml>

8.7 Recording of Materials

Presentations that are made in relation to course work - including lectures - cannot be recorded or copied without the permission of the presenter, whether the instructor, a student, or guest lecturer. Material recorded with permission is restricted to use for that course unless further permission is granted.

8.8 Resources

The Academic Calendars are the source of information about the University of Guelph's procedures, policies, and regulations that apply to undergraduate, graduate, and diploma programs.

Academic Calendars
<https://www.uoguelph.ca/academics/calendars>

8.9 Illness

Medical notes will not normally be required for singular instances of academic consideration, although students may be required to provide supporting documentation for multiple missed assessments or when involving a large part of a course (e.g.. final exam or major assignment).
