

Biology 3592A Course Outline Fall 2024 [K Hill Instructor]

1. Course Information

Course Information

Principles of Human Genetics 3592A Fall 2024

List of Prerequisites

Biochemistry 2280A

Biology 2581A/B [Introductory Genetics]

***Unless you have either the requisites for this course or written special permission from your Dean's Designate (Department/Program Counsellors and Science Academic Advisors) to enroll in it, you may be removed from this course and it will be deleted from your record. This decision may not be appealed. **You will receive no adjustment to your fees in the event that you are dropped from a course for failing to have the necessary prerequisites.**

2. Instructor Information

Instructors	Email*	Office	Contact	**Office Hours
Dr. Kathleen A. Hill, PhD (Course Coordinator; instructor)	khill22@uwo.ca	WSC333	Using the Help forum in Brightspace	TBA
TA Jordan VanderBurgt			Help forum in Brightspace	TBA
TA Joseph Butler			Help forum in Brightspace	TBA

*Students must use their **Western (@uwo.ca) email addresses** when contacting their instructors.

****Office hours format:** For questions related to course content, this course uses a combination of in-class review sessions and scheduled Zoom help sessions recorded and posted to OWL Brightspace. Other help resources used include OWL Brightspace Discussions where students can post questions that are answered by members of the teaching team. Discussions are monitored and attended to on weekdays by the instructor and the Teaching Assistants [TAs]. Questions related to individual student grades are directed to K Hill using khill22@uwo.ca.

3. Course Syllabus, Schedule, Delivery Mode



Syllabus, Required Readings, Lecture Schedule, and Tutorial Schedule are posted at the course OWL Brightspace site.

Weekly the resources posted include: Lecture slides; Lecture audio recording; tutorial slides; tutorial recording; Zoom help session recordings

Description of the objectives and content of the course:

PRINCIPLES TO PRACTICE

This course provides opportunities to learn fundamental discoveries in the scientific field of human genetics and genomics. We will study the history of the field of human genetics. We will construct and interpret inheritance patterns in human pedigrees. We will learn the nomenclature of human chromosome structure. We will use a collection of peer-reviewed journal articles as our guide to learning about interesting discoveries regarding the nature of the human genome and the dynamics of mutagenesis in the human genome. Tutorials will equip us with a working knowledge and understanding of key methods for the study of human genetic variation. The instructor and teaching assistants will provide teaching about their current research projects. Guest lecturers will present the latest findings from their research in clinical genetics, bioinformatics, genome evolution, cultural analytics, and research ethics. We will learn directly from academics, researchers, research ethics officers, clinician scientists and humanities researchers. Guests in our course include geneticists, bioinformaticians, social scientists and computer scientists. Assessments of knowledge and understanding are in the form of quizzes, a midterm test, a case study assignment, and the final exam.

The course description published in the Western Academic Calendar:

Genetic principles and their application to humans. Special attention will be directed to the genetic variation in our species, mutations, mechanisms of gene expression, and novel technologies and approaches to study the human genome.

Antirequisite(s): None **Prerequisite(s): Biochemistry 2280A; Biology 2581A/B.**

Corequisite(s): None Pre-or Corequisite(s): None

Extra Information: 2 lecture hours, 1 lecture/tutorial hour, 0.5 course

Course-level learning outcomes:

Upon completion of this course students will be familiar with the following key topics in current human genetics research:

- *De novo* postzygotic mutations
- Somatic and germline mosaicism
- Historical challenges and future projections for the field of human genetics
- Technologies, models, and methods in human genetics research
- Clever experimental designs in human genetics research [families, sibs, parent-progeny, twins]
- Anatomy of a human gene, human chromosome, and human genome
- Glossary of terms commonly used in human genetics
- Chromosomal anomalies
- Mutagens, Mutation types and mutational mechanisms
- Mobile elements in the human genome
- Communication in human genetics and sensitive language
- Case studies of human inherited disorders/diseases
- Ethical dilemmas in human genetics and genomics

- Human research ethics and the role of the research ethics office/officer

Learning Objectives and Course Outcomes:

On successful completion of this course students will be able to:

- Dissect and comprehend a primary research article, a perspectives article, and a review type article
 - Perform effective searches in reputable internet sources
 - Construct and interpret pedigree charts
 - Use and interpret human chromosome and karyotype nomenclature
 - Teach the anatomy of a human gene and The Central Dogma in Biology
 - Design experiments to detect different types of *de novo* mutations
 - Explain how mobile elements alter the human genome
 - Describe and illustrate common methods of DNA analysis
 - Join the conversation and identify ethical challenges presented by advances in human genetics/genomics research and the translation of new knowledge to the clinic and marketplace
 - Define and use sensitive language
 - Identify the science, technology, stakeholders, and issues in current bioethics challenges in human genetics/genomics
 - Understand, discuss, and debate current topics in bioethics associated with human genetic/genomic technologies
 - Describe genomic instability associated with inherited and acquired cancers
- Describe a broad spectrum of professions associated with human genetics

Lecture and Tutorial weekly meeting times

Delivery mode: Lectures and tutorials are typically presented in person on campus.

The teaching team will endeavour to record audio for lectures and tutorials and post that to Brightspace.

Blended Delivery refers to the *occasional* online teaching. In the event of planned or unanticipated online only lecture or tutorial sessions Brightspace will be used to post notice of these sessions and to send out email notification of these online only sessions.

The breakdown of the course topics and timeline is provided in an excel table format as a **lecture schedule** and a **tutorial schedule** posted to the course Brightspace.

Key Sessional Dates:

Western Undergraduate Classes begin: September 5, 2024

Non-instruction day: September 30 [no lectures this day] National Day for Truth and Reconciliation

Fall Reading Week: October 12 – 20; There are no lectures or tutorial this week

Midterm test: date/time/location TBA – awaiting test room confirmation

Midterm remake test: date/time/location TBA – awaiting test room confirmation

Classes end: December 6, 2024

Final Exam period: December 9 – 22, 2024

Final Exam: December date/time TBA; announced by the registrar

Special final exam: January date/time TBA; announced by the department/faculty

4. Course Materials

All course materials will be posted to OWL: <https://westernu.brightspace.com/>

The materials associated with the first lecture and the video recording of the first lecture are provided in the Brightspace course site in Week 1. The first lecture is a delivery of the course outline. *****This first lecture and first class are online only. There is no in-person class September 9.*****

There is no textbook for this course.

The **required readings** for the course are research publications and they are provided in Brightspace. Recommendations for optional supplemental resources will be provided in the lectures.

Students are responsible for checking the course OWL site (<https://westernu.brightspace.com/>) regularly for news and updates. This is the primary method by which information will be disseminated to all students in the class.

If students need assistance with the course OWL site, they can seek support on the [OWL Brightspace Help](#) page. Alternatively, they can contact the Western Technology Services Helpdesk. They can be contacted by phone at 519-661-3800 or ext. 83800.

Technical Requirements

Technical requirements for the course include stable internet connection, computer (working microphone and/or webcam are essential in the event of engagement with any Zoom class sessions). Internet and computer are used to access course content resources in Brightspace, to complete quizzes in Brightspace and to submit the course assignment to Brightspace. Planned use of Zoom is for special help sessions associated with the midterm test, course assignment and final exam.

A Statement regarding the use of Generative AI:

Coming soon as a separate document associated with the course assignment

5. Methods of Evaluation

Grading Scheme and Assessment Dates

The overall course grade will be calculated as listed below:

OWL Quizzes (#)	10%	Weekly tutorials; can be cumulative
*Midterm Test	25%	Specific Lectures and tutorials
*Midterm remake		Identical content to Midterm Test
Course Assignment	20%	as posted in OWL Brightspace
Final Exam	45%	Cumulative for all lectures and tutorials
Final Special Exam		Cumulative for all lectures and tutorials

Due Date or Writing Date

Date: posted in Brightspace
Date: Oct 30
Date: Nov 5
Date: November 19
Date: December TBA
Date: January TBA

**Awaiting test room location approval*

TBA = details are provided by the registrar and department/faculty

General information about missed coursework

Students must familiarize themselves with the *University Policy on Academic Consideration – Undergraduate Students in First Entry Programs* posted on the Academic Calendar: https://www.uwo.ca/univsec/pdf/academic_policies/appeals/academic_consideration_Sep24.pdf,

This policy does not apply to requests for **Academic Consideration** submitted for **attempted or completed work**, whether online or in person.

The policy also does not apply to students experiencing longer-term impacts on their academic responsibilities. These students should consult [Accessible Education](#).

For procedures on how to submit Academic Consideration Requests, please see the information posted on the Office of the Registrar's webpage:

https://registrar.uwo.ca/academics/academic_considerations/

All requests for Academic Consideration must be made within 48 hours after the assessment date or submission deadline.

All **Academic Consideration Requests** must include supporting documentation; however, recognizing that formal documentation may not be available in some extenuating circumstances, the policy allows students to make one Academic Consideration request **without supporting documentation** in this course. However, the following assessments are excluded from this, and therefore always require formal supporting documentation:

- **Examinations** scheduled during official examination periods (Defined by policy)
- **Midterm test and Remake Midterm test** (Designated by the instructor as the one assessment that always requires documentation when requesting Academic Consideration) (There is a remake test for those students with documentation and approved academic consideration for the Midterm test).

When a student *mistakenly* submits their one allowed Academic Consideration request **without supporting documentation** for the assessments listed above or those in the **Coursework with Assessment Flexibility** section below, the request cannot be recalled and reapplied. This privilege is forfeited.

Evaluation Scheme for Missed Assessments

A missed Midterm test is handled through Academic Accommodation Approval to permit the writing of the remake midterm test.

A missed Midterm test and a missed remake midterm test both with Academic Accommodation Approval is handled with the shift of the midterm 25% to the final exam making the final exam weight 70%.

A missed final exam is handled through the writing of the special final exam with Academic Accommodation Approval.

A missed final exam and a missed special final exam both with documentation and Academic Accommodation Approval is handled through the necessity to register for the course in the following year and write the final exam in that course offering during that examination period.

There are 10 quizzes in the course. The best 9 of 10 quizzes is used for the final grade. This gives flexibility and handles the event of a missed quiz. Each quiz is open for multiple days prior to the due date permitting students flexibility prior to the due date.

The assignment has a due date provided in the OWL Brightspace course site with the assignment instructions. Missing that due date is handled through a 48-hour period that is penalty-free. Missing the due date and the 48-hour penalty-free period results in penalty loss of 1% from the course grade for each 24-hour period after that.

The assignment submission cut-off date for the assignment is the last class of the term. That is the last date when submissions for the assignment will no longer be accepted even with Academic Consideration granted. This submission cut-off date is **ESPECIALLY IMPORTANT** for assessment where Academic Consideration can be granted without documentation (thus no end date). After this submission cut-off date, the Assignment weight will be used to reweigh the final exam. This is only the case for instances where Academic Consideration has been documented and granted for missing the due date.

The essential learning requirements of the course are the writing of the final examination [or special final examination for those with Academic Consideration documented and successfully granted].

When a student misses the Final Exam and Academic Consideration has been granted, they will be allowed to write the Special Examination (the name given by the University to a makeup Final Exam). See the Academic Calendar for details (under [Special Examinations](#)), especially for those who miss multiple final exams within one examination period.

Essential Learning Requirements

Even when Academic Considerations are granted for missed coursework, **the following are deemed essential to earn a passing grade for the course.**

Course-specific conditions related to essential learning objectives include:

- **a minimum passing grade [50%] on the final exam [or with academic approval to write the special examination] to ensure that students demonstrate sufficient mastery of the learning outcomes.**

In such cases, the consequences of not meeting the requirement (a grade of less than 50% on the final examination or final special examination) are a failing course grade.

For students who have not written the final examination or the special examination and have academic accommodation and permission to write the special examination the students register for the next offering of the course, in which case the student will receive a grade of Incomplete (INC) and their maximum course load may be reduced during the term in which they complete their course requirements.

Coursework with Assessment Flexibility

By policy, instructors may deny Academic Consideration requests for the following assessments with built-in flexibility:

Flexible Completion

Quizzes. Quizzes for this course are available for a multiple day period. This course has 10 quizzes, and the 9 quizzes with the highest marks are counted towards the final grade. Should extenuating circumstances arise, students do not need to request Academic Consideration for the first missed

quiz. Also, academic consideration requests will be denied for the first missed quiz. Academic Consideration requests may be granted when students miss 2 or more consecutive quizzes due to prolonged absences or situations preventing quiz completion [more than two weeks of consecutive absences]. The quiz grade may then be reweighted to the final exam for approved academic considerations for prolonged [multiple week] absences.

Deadline with a No-Late-Penalty Period

Course Assignment. Students are expected to submit the course assignment by the deadline listed. Should extenuating circumstances arise, students do not need to request Academic Consideration and they are permitted to submit their assignment up to 48 hours past the deadline without a late penalty. Should students submit their assignment beyond 48 hours past the deadline, a late penalty of 1% per 24-hour period will be applied. Academic Consideration requests may be granted only for extenuating circumstances that started before the deadline and lasted longer than the No-Late-Penalty Period (48 hours).

6. Additional Statements

Religious Accommodation

When conflicts with a religious holiday that requires an absence from the University or prohibits certain activities, students should request an accommodation for their absence in writing to the course instructor and/or the Academic Advising office of their Faculty of Registration. This notice should be made as early as possible but not later than two weeks prior to the writing or the examination (or one week prior to the writing of the test).

Please visit the Diversity Calendars posted on our university's EDID website for the recognized religious holidays: <https://www.edi.uwo.ca>.

Accommodation Policies

Students with disabilities are encouraged to contact Accessible Education, which provides recommendations for accommodation based on medical documentation or psychological and cognitive testing. The policy on Academic Accommodation for Students with Disabilities can be found at:

https://www.uwo.ca/univsec/pdf/academic_policies/appeals/Academic_Accommodation_disabilities.pdf.

Academic Policies

The website for Registrar Services is <https://www.registrar.uwo.ca/>.

In accordance with policy,

https://www.uwo.ca/univsec/pdf/policies_procedures/section1/mapp113.pdf,

the centrally administered e-mail account provided to students will be considered the individual's official university e-mail address. It is the responsibility of the account holder to ensure that e-mail received from the University at their official university address is attended to in a timely manner.

No electronic devices are permitted on tests and exams.

Scholastic offences are taken seriously, and students are directed to read the appropriate policy, specifically, the definition of what constitutes a Scholastic Offence, at the following Web site:

https://www.uwo.ca/univsec/pdf/academic_policies/appeals/scholastic_discipline_undergrad.pdf.

All required papers may be subject to submission for textual similarity review to the commercial plagiarism detection software under license to the University for the detection of plagiarism. All papers submitted for such checking will be included as source documents in the reference database for the purpose of detecting plagiarism of papers subsequently submitted to the system. Use of the service is subject to the licensing agreement, currently between The University of Western Ontario and Turnitin.com (<http://www.turnitin.com>).

Computer-marked multiple-choice tests and exams may be subject to submission for similarity review by software that will check for unusual coincidences in answer patterns that may indicate cheating.

Support Services

Please visit the Science & Basic Medical Sciences Academic Advising webpage for information on adding/dropping courses, academic considerations for absences, appeals, exam conflicts, and many other academic-related matters: <https://www.uwo.ca/sci/counselling/>.

Students who are in emotional/mental distress should refer to Mental Health@Western (<https://uwo.ca/health/>) for a complete list of options about how to obtain help.

Western is committed to reducing incidents of gender-based and sexual violence and providing compassionate support to anyone who has gone through these traumatic events. If you have experienced sexual or gender-based violence (either recently or in the past), you will find information about support services for survivors, including emergency contacts at

https://www.uwo.ca/health/student_support/survivor_support/get-help.html.

To connect with a case manager or set up an appointment, please contact support@uwo.ca.

Please contact the course instructor if you require lecture or printed material in an alternate format or if any other arrangements can make this course more accessible to you. You may also wish to contact Accessible Education at

http://academicsupport.uwo.ca/accessible_education/index.html

if you have any questions regarding accommodations.

Learning-skills counsellors at Learning Development and Success (<https://learning.uwo.ca>) are ready to help you improve your learning skills. They offer presentations on strategies for improving time management, multiple-choice exam preparation/writing, textbook reading, and more. Individual support is offered throughout the Fall/Winter terms in the drop-in Learning Help Centre, and year-round through individual counselling.

Western University is committed to a thriving campus as we deliver our courses in the mixed model of both virtual and face-to-face formats. We encourage you to check out the Digital Student Experience website to manage your academics and well-being: <https://www.uwo.ca/se/digital/>.

Additional student-run support services are offered by the USC, <https://westernusc.ca/services/>.

Helpful online resources:

Genomes [2nd Ed] available online: <https://www.ncbi.nlm.nih.gov/books/NBK21128/>

Relevant chapters recommended: Chapters 1, 2, 4-8, 14.

Genes and Diseases: <https://www.ncbi.nlm.nih.gov/books/NBK22183/>

Genetics for Surgeons: <https://www.ncbi.nlm.nih.gov/books/NBK7524/>

There are several online texts that contain helpful information and explanations for the fundamental principles taught in this course. The online books have a nice search tool so you can rapidly find text relevant to the course topics. I recommend using this search tool for concepts mentioned in the readings and lectures to help find definitions and helpful figures to explain basic concepts and fundamental principles. If the links in this course outline fail to directly open, please copy and paste the link into your internet browser.

Help with Writing - Writing Support Centre – Student Development Centre

Home page: <https://writing.uwo.ca/> - note Zoom appointments can be made! And there is online assistance! This site also has Writing Resources: Writing Support Handouts in Video, Word, and pdf format ready for download

There is Online Writing Help.

There are Weldon Drop-in Services.

There are One-to-one Appointments with a writing advisor.

Professionalism & Privacy:

Western students are expected to follow the Student Code of Conduct.

Additionally, the following expectations and professional conduct apply to this course:

- Students are expected to follow online etiquette expectations provided on OWL
- All course materials created by the instructor(s) are copyrighted and cannot be sold/shared
- Recordings are not permitted (audio or video) without explicit permission
- Permitted recordings are not to be distributed
- Students will be expected to take an academic integrity pledge before some assessments
- All recorded sessions will remain within the course site or unlisted if streamed

General Online Engagement Etiquette –

- Some components of this course will involve online interactions. To ensure the best experience for both you and your classmates, please honour the following rules of etiquette:
 - “arrive” to class on time
 - use your computer and/or laptop if possible (as opposed to a cell phone or tablet)
 - ensure that you are in a private location to protect the confidentiality of discussions in the event that a class discussion deals with sensitive or personal material
 - to minimize background noise, mute your microphone for the entire class until you are invited to speak, unless directed otherwise
 - [suggested for classes larger than 30 students] In order to give us optimum bandwidth and web quality, turn off your video camera for the entire class unless you are invited to speak
 - [suggested for cases where video is used] please be prepared to turn your video camera off at the instructor’s request if the internet connection becomes unstable
 - unless invited by your instructor, do not share your screen in the meeting
- The course instructor will act as moderator for the class and will deal with any questions from participants. To participate please consider the following: If you wish to speak, use the “raise hand” function and wait for the instructor to acknowledge you before beginning your comment or question.
- Please remember to unmute your microphone and turn on your video camera before speaking.
- Self-identify when speaking.
- Please remember to mute your mic and turn off your video camera after speaking (unless directed otherwise).

General considerations of “netiquette”:

- Keep in mind the different cultural and linguistic backgrounds of the students in the course.
- Be courteous toward the instructor, your colleagues, and authors whose work you are discussing.
- Be respectful of the diversity of viewpoints that you will encounter in the class and in your readings. The exchange of diverse ideas and opinions is part of the scholarly environment. “Flaming” is never appropriate.
- Be professional and scholarly in all online postings. Use proper grammar and spelling. Cite the ideas of others appropriately.
- Note that disruptive behaviour of any type during online classes, including inappropriate use of the chat function, is unacceptable. Students found guilty of Zoom-bombing a class or of other serious online offenses may be subject to disciplinary measures under the Code of Student Conduct.