

COURSE OUTLINES (SYLLABI) FOR UNDERGRADUATE COURSES

1. Course Information

Course Information

Course name and number: 4560B Human Molecular Genetics

academic term: 2023 Winter term,

Lectures: Tuesday & Thursday 8:30-9:30 NCB-293

List of Prerequisites

Unless you have either the requisites for this course or written special permission from your Dean 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.

The prerequisite for this class is Biology 3338a.

2. Instructor Information

Full name, title and appropriate contact information of the instructor: Dr. Miodrag (Mike) Grbic, Associate Professor

e-mail: mgrbic@uwo.ca

Students must use their Western (@uwo.ca) email addresses when contacting their instructors. [insert other contact policies here]

Office hours: flexible, on demand

Format scheduled: in person/Zoom online

3. Course Syllabus, Schedule, Delivery Mode

In this course we will focus on human genome, molecular basis of human genetic disease, genetics of

polygenic diseases and traits, genome evolution, non-coding RNA, chromatin re-modelling, molecular aspects of cancer genetics, epigenetics, stem cell biology particularly relevant to human disease - including the application of iPS cells, therapy of genetic disease, pharmacogenetics, biochemical genetics, advances in gene mapping, and functional genomics. Studies in model organisms are included where they show novel insights into disease.

Course-level learning outcomes:

Student experience

By the end of this course, students will have:

- a) Will have a broad knowledge of approaches, problems and tools in Human Molecular biology studies focusing on genome and genomic/genetic approaches
- b) Will have knowledge on genetics of human diseases and novel approaches of disease detection, diagnosis and treatments
- c) Will be able to read and critically analyse primary literature and apply knowledge to new situations

Assessment

The course utilizes two mid-term exams and final exam as an assessment tool. The mid-term exams are testing knowledge and understanding of the genetics of human disorders and human basic functions. The format of the exam is problem solving and interpretation of the data. The final assignment is placing students in role of scientist analyzing key primary literature paper and applying knowledge acquired on the course for solving novel problems not presented on the course. The format of exams is tailored not to test student's stress response but to provide environment stimulating creative and critical thinking.

Lectures: Tuesday & Thursday 8:30-9:30 (virtual synchronous and virtual asynchronous);

Course plan (see also course planning description)

Lectures (pages given based on Strachan 4th edition):

- 1) Tuesday Jan. 10. Human Molecular Biology: Introduction:
 - Why is human genetics important?
- 2) Thursday Jan. 12. Human genome project and organization of the human genome I p.256-294
- 3) Tuesday Jan. 17. Human genome project and organization of the human genome II p.256-294

- 4) Thursday Jan. 19. Studying the gene function in post genomic era p. 382-404
- 5) Tuesday Jan. 24. Comparative Genomics and Evolution of Human genome I p. 297-339
- 6) Thursday Jan. 26. Comparative Genomics and Evolution of Human genome II p. 297-339
- 7) Tuesday Jan. 31. Human gene expression I p. 347-378
- 8) Thursday Feb. 02. Human gene expression II p.347-378
- 9) Tuesday Feb. 7. Human Genetic variability and its consequences I p. 406-438
- 10) Thursday Feb. 9. Human Genetic variability and its consequences II p. 406-438
- 11) Tuesday Feb. 14. Identification of human disease genes and susceptibility factors I p. 498-535
- 12) Thursday Feb. 16. In class first mid-term to be confirmed (30% grade before March 1)
 - Tuesday Feb. 21. Reading week
 - Thursday Feb. 23. Reading week
- 13) Tuesday Feb. 28. Identification of human disease genes and susceptibility factors II p. 498-535
- 14) Thursday Mar. 02. Pharmacogenetics, personalized medicine and population screening I p. 606-637.
- Tuesday Mar. 07. Pharmacogenetics, personalized medicine and population screening II p. 606-637.
- 16) Thursday Mar. 9. Cancer genetics I p. 538-567.
- 17) Tuesday Mar. 14. Cancer genetics II p. 538-567.
- 18) Thursday Mar. 16. Genetic approaches to treating disease I p. 678-716
- 19) Tuesday Mar. 21. Genetic approaches to treating disease II p. 678-716
- 20) Thursday Mar. 23. In class second midterm to be confirmed (30% grade; 15 days before end of term)

- Tuesday Mar. 28. Genetic manipulation of animals for modeling diseases and investigating gene function I p. 640-672
- 22) Thursday Mar. 30. Genetic manipulation of animals for modeling diseases and investigating gene function II p. 640-672
- 23) Tuesday April. 4. Genetic testing of individuals I p. 569-601
- 24) Thursday April. 6. Life style and health.
- 25) Tuesday April. 11. . Life style and health

Final assignment due: April 17/th (to be deposited to course dropbox) Will be placed on OWL in January

Evaluation: 2 in class midterms (30% each), Final assignment 40%

Key Sessional Dates listed in the 'Course Planning Info' document:

Classes begin: January 9th Reading Week: February 21-26

Classes end: April 13

4. Course Materials

Human Molecular Genetics / Edition 4 or 5 (Tom Strachan & Andrew Read) and selected papers from primary literature

Students should check OWL (http://owl.uwo.ca) on a regular basis for news and updates. This is the primary method by which information will be disseminated to all students in the class. Students are responsible for checking OWL on a regular basis.

All course material will be posted to OWL: http://owl.uwo.ca.

If students need assistance, they can seek support on the OWL 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

Stable internet connection, computer with working microphone and webcam

5. Methods of Evaluation

A statement of the methods by which student performance will be evaluated and the weight of each, including an exact timetable and schedule of assignments (due date), is required.

The overall course grade will be calculated as listed below:

First Midterm 30% Second Midterm 30% Final assignment 40%

The course utilizes two mid-term exams and final exam as an assessment tool. The mid-term exams are testing knowledge and understanding of the genetics of human disorders and human basic functions. The format of the exam is problem solving and interpretation of the data. The final asignment is placing students in role of scientist analyzing key primary literature paper and applying knowledge acquired on the course for solving novel problems not presented on the course. The format of exams is tailored not to test student's stress response but to provide environment stimulating creative and critical thinking.

To minimize the number of conflicts between tests that could occur if instructors are given free rein to schedule their own tests, the OOR is developing a mechanism to request times for midterm tests. In addition, central management is necessary to ensure that capacity for proctored online exams is not exceeded. Instructors are advised <u>not</u> to specify dates and times for any midterms tests until those times have been verified.

For Fall/Winter of 2022-23, assignments may be due <u>later than the last day of scheduled classes</u> under the following conditions:

- when the class has no examination or other assessment during the exam period,
- when the assignment could be reasonably completed in 12 hours or less, and
- when the assignment can be adequately completed by responses totaling 2500 words or less (1500 words or less for a course numbered 1999 or lower).

When assignments satisfying these conditions are set during the examination period, they should be due no earlier than the tenth day of the examination period.

Final assignment due date: April 18th 2023

Accommodated Evaluations

Conditions might include (i) minimum attendance at lectures, tutorials or laboratories, (ii) minimum overall grade on laboratory, tutorial or essay components of the methods of evaluation, or (iii) minimum required grades on a final exam, to name a few.

Note: The requirement of minimum 'Attendance' may be poorly defined for an asynchronous online course. Instead, use a minimum 'level of engagement' instead (e.g., forum posting).

If permission to waive the requirement that students receive evaluation on work totaling 15% of their final grade at least three days prior to the deadline for withdrawal without academic penalty has been obtained from the Dean's Office, a statement to this effect must be made.

Biology 4560B is an essay course, and therefore, failure (potentially as a result of plagiarism) or noncompletion of the essay component of the course will result in failure of the course and awarding a final course mark of 45%.

If you are unable to meet a course requirement due to illness or other serious circumstances, you must provide valid medical or supporting documentation to the Academic Counselling Office of your home faculty as soon as possible. If you are a Science student, the Academic Counselling Office of the Faculty of Science is located in WSC 140, and can be contacted at 519- 661-3040 or scibmsac@uwo.ca. Their website is http://www.uwo.ca/sci/undergrad/academic counselling/index.html. A student requiring academic accommodation due to illness must use the Student Medical Certificate (https://studentservices.uwo.ca/secure/medical_document.pdf) when visiting an off-campus medical facility. For further information, please consult the university's medical illness policy at http://www.uwo.ca/univsec/pdf/academic_policies/appeals/accomm odation_medical.pdf.

The late penalty for assignments is as follows. Essay is out of 20 marks; 4 marks will be deducted from the mark received for the first day late, and 2 marks for each subsequent day late. In addition, noncompletion or failure of the essay will result in automatic failure of the course (see Senate rules for essay courses). Therefore, plagiarism results in failure of the course.

6. Accommodation and Accessibility

Accommodation Policies

Students with disabilities work with Accessible Education (formerly SSD) which provides recommendations for accommodation based on medical documentation or psychological and cognitive testing. The Academic Accommodation for Students with Disabilities policy can be found at: https://www.uwo.ca/univsec/pdf/academic_policies/appeals/Academic Accommodation_disabilities.pdf

Academic Consideration for Student Absence

Students will have up to two (2) opportunities during the regular academic year to use an on-line portal to self-report an absence during the semester, provided the following conditions are met: the absence is no more than 48 hours in duration, and the assessment for which consideration is being sought is worth 30% or less of the student's final grade. Students are expected to contact their instructors within 24 hours of the end of the period of the self-reported absence, unless noted on the syllabus. Students are not able to use the self-reporting option in the following circumstances:

- for exams scheduled by the Office of the Registrar (e.g., December and April exams)
- absence of a duration greater than 48 hours,
- assessments worth more than 30% of the student's final grade,
- if a student has already used the self-reporting portal twice during the academic year

If the conditions for a Self-Reported Absence are *not* met, students will need to provide a Student Medical Certificate if the absence is medical, or provide appropriate documentation if there are compassionate grounds for the absence in question. Students are encouraged to contact their Faculty academic counselling office to obtain more information about the relevant documentation.

Students should also note that individual instructors are not permitted to receive documentation directly from a student, whether in support of an application for consideration on medical grounds, or for other reasons. All documentation required for absences that are not covered by the Self-Reported Absence Policy must be submitted to the Academic Counselling office of a student's Home Faculty.

For policy on Academic Consideration for Student Absences - Undergraduate Students in First Entry Programs, see:

https://www.uwo.ca/univsec/pdf/academic_policies/appeals/Academic_Consideration_for_absences.pdf and for the Student Medical Certificate (SMC), see:

http://www.uwo.ca/univsec/pdf/academic_policies/appeals/medicalform.pdf

Religious Accommodation

Students should consult the University's list of recognized religious holidays, and should give reasonable notice in writing, prior to the holiday, to the Instructor and an Academic Counsellor if their course requirements will be affected by a religious observance. Additional information is given in the Western Multicultural Calendar:

https://multiculturalcalendar.com/ecal/index.php?s=c-univwo

You may also be eligible to write the Special Exam if you are in a "Multiple Exam Situation" (see http://www.registrar.uwo.ca/examinations/exam_schedule.html).

[optional] If a student fails to write a scheduled Special Examination, the date of the next Special Examination (if granted) normally will be the scheduled date for the final exam the next time this course is offered. The maximum course load for that term will be reduced by the credit of the course(s) for which the final examination has been deferred. See Academic Calendar for details (under Special Examinations).

7. Academic Policies

The website for Registrarial Services is http://www.registrar.uwo.ca.

In accordance with policy, http://www.uwo.ca/its/identity/activatenonstudent.html, 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 his/her official university address is attended to in a timely manner.

Contingency plan for an in-person class pivoting to 100% online learning

In the event of a COVID-19 resurgence during the course that necessitates the course delivery moving away from face-to-face interaction, all remaining course content will be delivered entirely online, either synchronously (i.e., at the times indicated in the timetable) or asynchronously (e.g., posted on OWL for students to view at their convenience). The grading scheme will **not** change. Any remaining assessments will also be conducted online as determined by the course instructor.

All of the remote learning sessions for this course will be recorded. The data captured during these recordings may include your image, voice recordings, chat logs and personal identifiers (name displayed on the screen). The recordings will be used for educational purposes related to this course, including evaluations. The recordings may be disclosed to other individuals participating in the course for their private or group study purposes. Please contact the instructor if you have any concerns related to session recordings.

Participants in this course are not permitted to record the sessions, except where recording is an approved accommodation, or the participant has the prior written permission of the instructor.

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: http://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.

8. Support Services

Please visit the Science & Basic Medical Sciences Academic Counselling webpage for information on add/drop courses, academic considerations for absences, appeals, exam conflicts, and many other academic related matters: https://www.uwo.ca/sci/counselling/

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 Student Accessibility Services (SAS) at (519) 661-2147 if you have any questions regarding accommodations.

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/.

Learning-skills counsellors at the Student Development Centre (http://www.sdc.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.

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

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