Western Science

Department of Biology

Human Molecular Genetics
Biology (4560B)
Course Outline/Syllabus Winter 2025

1. Course Information

Course Information
Human Molecular Genetics**,** Biology (4560B);

List of Prerequisites: Bio 3592, Bio 3596 and any bio course at level 3000 or above
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

Professor	**Email**	**Office**	**Phone**	**Office Hours**
Miodrag Grbic	mgrbic@uwo.ca	WSC 342	519-661-2111 X 67776	9:30-10:30 Tu Thu in person or zoom and/or other time on demand
GTAs:				

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

3. Course Syllabus, Schedule, Delivery Mode

Course Description: In this course, we will focus on the human genome, the molecular basis of human genetic disease, genetics of polygenic diseases and traits, genome evolution, non-coding RNA, chromatin re-modeling, 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.

Learning Outcomes and Objectives: 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 gain knowledge on the genetics of human diseases and novel approaches to disease detection, diagnosis, and treatments
c) Will be able to read and critically analyze primary literature and apply knowledge to new situations

Contingency Plan: This course will be delivered in-person, however in the unlikely event of any university-declared emergency, some or all of this course may be required to be delivered online, either synchronously or asynchronously. ***The grading scheme will not change***. Any assessments affected will be conducted online as determined by the professor.

Classes begin: January 6, 2025
Spring Reading Week: February 17 – 21, 2025
Classes end: April 4, 2025
Exam period: April 7 – 30, 2025

4. Course Materials

Course plan (see also course planning description)

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

1) Tuesday Jan. 7. Human Molecular Biology: Introduction :

   Why is human genetics important?

2) Thursday Jan. 9. Human genome project and organization of the human genome I p.256-294

3) Tuesday Jan. 14. Human genome project and organization of the human genome II p.256-294

4) Thursday Jan. 16. Studying the gene function in post genomic era p. 382-404

5) Tuesday Jan. 21. Comparative Genomics and Evolution of Human genome I p. 297-339

6) Thursday Jan. 23. Comparative Genomics and Evolution of Human genome II p. 297-339

7) Tuesday Jan. 28. Human gene expression I p. 347-378

8) Thursday Feb. 30. Human gene expression II p.347-378

9) Tuesday Feb. 4. Human Genetic variability and its consequences I p. 406-438

10) Thursday Feb. 6. Human Genetic variability and its consequences II p. 406-438

11) Tuesday Feb. 11. Identification of human disease genes and susceptibility factors I p. 498-535

12) Thursday Feb. 13. In class first mid-term to be confirmed (30% grade before March 1)
All course material will be posted to OWL: https://westernu.brightspace.com/

Tuesday Feb. 15. Reading week

13) Tuesday Feb. 25. Identification of human disease genes and susceptibility factors II p. 498-535

14) Thursday Feb. 27. Pharmacogenetics, personalized medicine and population screening I p. 606-637.

15) Tuesday Mar. 4. Pharmacogenetics, personalized medicine and population screening II p. 606-637.

16) Thursday Mar. 6. Cancer genetics I p. 538-567.

17) Tuesday Mar. 11. Cancer genetics II p. 538-567.

18) Thursday Mar. 13. Genetic approaches to treating disease I p. 678-716

19) Tuesday Mar. 18. Genetic approaches to treating disease II p. 678-716

20) Thursday Mar. 20. In class second midterm to be confirmed (30% grade; 15 days before end of term)

21) Tuesday Mar. 25. Genetic manipulation of animals for modeling diseases and investigating gene function I p. 640-672

22) Thursday Mar. 27. Genetic manipulation of animals for modeling diseases and investigating gene function II p. 640-672

23) Tuesday April. 1. Genetic testing of individuals I p. 569-601

24) Thursday April. 3. Life style and health.

Final assignment due: April 13th midnight (to be deposited to course dropbox)

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

Required Equipment:

Stable internet connection

Laptop or computer with webcam

Working microphone

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

All course documents and information (syllabus, lecture files, instructional documents, required readings, etc.) **will be posted to OWL and students are responsible for checking the course OWL site (http://owl.uwo.ca) on a regular basis for updates.** If students need assistance with the course OWL site, they can seek support on the OWL Help page. Alternatively, they can contact the Western Technology Services Helpdesk, which can be contacted by phone at 519-661-3800 or ext. 83800.

Personal Response Systems ("Clickers") are not used in this course.

5. Methods of Evaluation

Grading Scheme and Assessment Dates

The overall course grade will be calculated as:

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

The course utilizes two mid-term exams and a final assignment 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 the role of a scientist analyzing key primary literature paper and applying knowledge acquired in the course for solving novel problems not presented in the course. The format of exams is tailored not to test student's stress response but to provide an environment stimulating creative and critical thinking.

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)

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
Make-up exams will be organized upon request based on student service approval of Academic Consideration within a week after in class midterm exam

When a student misses the Final Exam and their 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.

- a minimum grade on the final assignment (or a minimum average on the midterm test(s) and final assignment) to ensure that students demonstrate sufficient mastery of the learning outcomes

[If applicable] **Coursework with Assessment Flexibility**
By policy, instructors may deny Academic Consideration requests for the following assessments with built-in flexibility:

Deadline with a No-Late-Penalty Period
Assignments. Students are expected to submit each of the the final 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 24 hours past the deadline without a late penalty. Should students submit their assessment beyond 24 hours past the deadline, a late penalty of 10% per day 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 (24 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 will be 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).

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/.