

Pathology and Laboratory Medicine
Genetics and Epigenetics of Human Disease, Pathology 4600B

Tuesday, 5:30-7:30pm

This course takes place at Western University, which is located on the traditional territories of the Anishinaabek, Haudenosaunee, Lūnaapéewak, and Chonnonton Nations, on lands connected with the London Township and Sombra Treaties of 1796 and the Dish with One Spoon Covenant Wampum.

Students who are in emotional and/or mental distress should refer to <https://www.uwo.ca/health/> for a complete list of options about how to obtain help.

1. Technical Requirements:



Stable internet connection



Laptop or computer

2. Important Dates:

Classes Begin	Reading Week	Classes End	Study day(s)	Exam Period
January 5	February 14–22	April 9	April 10, 11	April 12–30

January 13, 2026: Last day to add or drop a Winter 12-week course

March 30, 2026: Last day to withdraw from a Winter 12-week course without academic penalty

3. Contact Information

Course Coordinator	Contact Information
Fred Dick	

Instructor(s) or Teaching Assistant(s)	Contact Information

4. Course Description and Design

Delivery Mode: in-person

This course encompasses the origins of genetic and epigenetics changes found in patients, manifestations of these in disease, and treatment approaches. Most disease scenarios will focus on inheritance of developmental and degenerative diseases. Mouse models will highlight causative changes as a complement to the analysis of human clinical examples.

Prerequisite(s): Biology 2581A/B; Pathology 3500

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.

Timetabled Sessions

Component	Date(s)	Time
In-person lectures	Tuesdays	5:30-7:20pm

- Students should familiarize themselves with lecture materials and readings prior to sessions to ensure they are prepared to participate in discussions.
- Students should bring a laptop, tablet, or device that they are comfortable typing with and searching the internet as most classes will include skill building exercises for searching genetic databases.
- Attendance at sessions is required

All course material will be posted to OWL Brightspace: <https://westernu.brightspace.com/d2l/login>. Any changes will be indicated on the OWL Brightspace site and discussed with the class.

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

Current versions of all popular browsers (e.g., Safari, Chrome, Edge, Firefox) are supported with OWL Brightspace; what is most important is that you update your browser frequently to ensure it is current. All JavaScript and cookies should be enabled.

5. Learning Outcomes

Upon successful completion of this course, students will be able to:

- Be able to describe molecular origins of genetic and epigenetic changes associated with diseases, such as cancer, and developmental anomalies in humans.
- Be able to describe and compare modern molecular genetic diagnostic methods.
- Be able to explain how DNA sequence aberrations resulted in selected examples of human disease conditions.
- Be able to explain how epigenetic alterations resulted in selected examples of human disease conditions.
- Be able to distinguish different clinical trials and therapeutic repurposing in the management of select examples of diseases caused by DNA sequence aberrations and epigenetic alterations.
- Become familiar with the use and interpretation of information from human genetic databases.

6. Course Content and Schedule

Week	Dates	Topic	Instructor
1	Jan 6	1) Course overview/Nuclear structure 2) Mitosis/Genome browsers	Dick
2	Jan 13	3) Meiosis/Replication of DNA 4) DNA damage and repair/OMIM database	Dick
3	Jan 20	5) Epigenetic regulation 6) Repetitive genomic regions/ cBioPortal	Dick
4	Jan 27	7) Neurodegenerative, unstable repeat disorders 8) mitochondrial disorders and resources	Schenkel
5	Feb 3	9) Hereditary cancer syndromes and mutations in DNA repair and cell cycle genes 10) Tools in diagnostics in cancer: somatic x hereditary testing	Lalonde
6	Feb 10	11) Imprinting disorders 12) Cytogenetic diagnostics and databases	Meybodi
	Feb 16–22	Reading Week	
7	Feb 24	13) Condensinopathies and Cohesinopathies 14) Genetic Association/genetic risk alleles	Dick
8	Mar 3	15) Clinical trial basics and the beginnings of molecular targeted therapy 16) Approaches to treating genetic deficiencies	Dick
9	Mar 10	17) Review class	Dick
10	Mar 17	In class Quiz	N/A
11	Mar 24	18) Genetic engineering in sickle cell anemia 19) Anti-sense oligonucleotide therapeutics as treatments for genetic disease	Dick
12	Mar 31	20) Synthetic Lethality 21) Epigenetics in Human Complex Diseases	Schenkel and Lalonde
13	Apr 7	22/23) Discussion/Review	Dick

7. Participation and Engagement

- Students are expected to participate and engage with content as much as possible. This is a small course and there will plenty of opportunity for questions and discussion.
- Students are encouraged to use the question and discussion period that follows each weekly course meeting at 7:20pm.
- Students can participate by interacting in the forums with their peers and instructors

8. Assessment and Evaluation

Below is the evaluation breakdown for the course. Any deviations will be communicated.

Assessment	Format	Weighting	Due Date	Flexibility
Assignment #1	Online submission of 'genetics in the news' and personal statement.	6	Jan. 8	48-hour no late penalty, re-weighting possible

Assignment #2	Online submission of 'database focused problem'.	6	Jan. 22	48-hour no late penalty, re-weighting possible
Assignment #3	Online submission of 'Clinical interpretation of molecular variants' with explanations.	6	Feb. 5	48-hour no late penalty, re-weighting possible
Take home quiz	Online timed quiz testing background and acquisition of skills	15	Feb. 11-13	48-hour no late penalty, re-weighting possible
Assignment #4	Online submission of 'Clinical interpretation of chromosomal variants' with explanations.	6	Feb. 26	48-hour no late penalty, re-weighting possible
In class quiz	Short answer and data interpretation based problem solving questions.	20	Mar. 17	N/A, designated assessment
Assignment #5	Online submission of 'Targeted therapeutics clinical trial' with explanations.	6	Mar. 26	48-hour no late penalty, re-weighting possible
Final exam	Short answer and data interpretation based problem solving questions.	35	TBD	N/A, designated assessment

Designated Assessment: Instructors are permitted to designate one assessment per course per term as requiring supporting documentation to receive academic consideration. See below for information on academic consideration policy and missed course work. The following assessment has been designated as requiring supporting documentation:

- In class Quiz AND documentation is DUE March 16th

Information about flexibility in assessment

- All online assignments and online quiz in this course will have an automatic grace period where submission can occur up until 48 hours past the deadline without penalty. Special permission is not required to utilize this grace period.
- Assignments or quizzes that are ultimately missed will lead to re-weighting of the final exam as a means to resolve grading of these items.
- The in class quiz and final exams will have one make-up time slot offered (date to be determined).

General information about assessments

- All online assignments and quizzes are due at 8:00 PM EST unless otherwise specified.
- Students are responsible for ensuring that the correct file version is uploaded; incorrect submissions including corrupt files could be subject to a 0 grade.
- Rubrics will be used to evaluate assessments and will be posted with the instructions
- After an assessment is returned, students should wait 24 hours to digest feedback before contacting their evaluator; to ensure a timely response, reach out within 7 days
- Any grade appeals on assignments, quizzes, or midterms must be received within 3 weeks of the grade being posted.

- 33% of your course grade will be evaluated and returned by the end of reading week, February 22nd.

The table below outlines the University-wide grade descriptors.

A+	90-100	One could scarcely expect better from a student at this level
A	80-89	Superior work which is clearly above average
B	70-79	Good work, meeting all requirements, and eminently satisfactory
C	60-69	Competent work, meeting requirements
D	50-59	Fair work, minimally acceptable
F	below 50	Fail

Information about late or missed assessments:

- All online assignments and quiz in this course will have an automatic ‘no penalty’ period where submission can occur up until 48 hours past the deadline. Special permission is not required to utilize this grace period.
- Assignments or quizzes that are ultimately missed will lead to re-weighting of the final exam to make up these items.
- The mid-term and final exam are essential to pass the course. If these elements are missed the grade will be INC, and the student will complete it the next time the course is offered.
- If a make-up exam is missed without documentation, the student will receive an INC and complete the task next time the course is offered.

INC (Incomplete Standing): If a student has been approved by the Academic Advising Office (in consultation with the instructor/department) to complete term work at a later date, an INC will be assigned, which could impact program progression. Students with INC will have their course load in subsequent terms reduced to allow them to complete outstanding course work. Students may request permission from Academic Advising to carry a full course load for the term the incomplete course work is scheduled.

SPC (Special examination): If a student has been approved by the Academic Advising Office to write a Special Examination and the final exam is the only outstanding course component, an SPC will be assigned. If the class has a makeup exam, the student is expected to write the makeup exam. If the class doesn't have a makeup for the final exam or the student misses the makeup for the final exam for reasons approved by the Academic Advising Office, the student will write the exam the next time the course is offered, which could impact program progression. Outstanding SPCs will reduce the course load for the term the exam is deferred as outlined in [Definitions of Types of Examinations](#) policy.

9. Communication

- Students should check the OWL Brightspace on a routine basis, multiple times per week.
- Students should contact their instructor(s) and teaching assistant(s) using email addresses.
- Emails will be monitored [daily]; students will receive a response in 24–48 hours
- This course will use discussions on Brightspace, but the primary form of question asking and answering will be in class. Posted questions may be answered verbally in class or distributed in course announcements so that all students will have access to them.

10. Office Hours

- Since instructors are located at the Victoria hospital site, the best opportunity to ask questions live is at the conclusion of lectures and during the break time between lectures.
- Individual or group meetings on Zoom can be arranged. Office hours will be [booked or drop in]

11. Course Materials

- All resources will be posted on OWL Brightspace
- No required textbook

12. Professionalism & Privacy

Western students are expected to follow the [Student Code of Conduct](#). Additionally, the following expectations and professional conduct apply to this course:

- All course materials created by the instructor(s) are copyrighted and cannot be sold/shared/upload (e.g., Must Knows Facebook group, Course Hero, Chegg, ChatGPT, etc.)
- 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 assessments.

Western is committed to providing a learning and working environment that is free of harassment and discrimination. All **students**, staff, and faculty have a role in this commitment and have a responsibility to ensure and promote a safe and respectful learning and working environment. Relevant policies include Western's [Non-Discrimination/Harassment Policy \(M.A.P.P. 1.35\)](#) and [Non-Discrimination/Harassment Policy – Administrative Procedures \(M.A.P.P. 1.35\)](#). Any **student**, staff, or faculty member who experiences or witnesses' behaviour that may be harassment or discrimination **must report the behaviour** to the Western's [Human Rights Office](#). Harassment and discrimination can be human rights-based, which is also known as EDI-based, (sexism, racism, transphobia, homophobia, islamophobia, xenophobia, antisemitism, and ableism) or non-human rights-based (personal harassment or workplace harassment).

13. How to Be Successful in this Class

Students enrolled in this class should understand the level of autonomy and self-discipline required to be successful.

1. Invest in a planner or application to keep track of your courses. Populate all your deadlines at the start of the term and schedule your time throughout the course.
2. Make it a habit to log onto OWL Brightspace to ensure you have seen everything posted to help you succeed in this class.
3. Follow checklists created on OWL Brightspace or create your own to help you stay on track.
4. Take notes as you go through the lesson material. Keeping handwritten notes or even notes on a regular Word document will help you learn more effectively than just reading or watching the videos.
5. Connect with others. Try forming a study group and try meeting on a weekly basis for study and peer support, this can be particularly important for building the skills needed for assignments and tests.
6. Do not be afraid to ask questions. If you are struggling with a topic, check the online discussion boards or contact your instructor(s) and or teaching assistant(s).
7. Reward yourself for successes. It seems easier to motivate ourselves knowing that there is something waiting for us at the end of the task.

14. Western Academic Policies/Procedures and Statements

A. Absence from Course Commitments

[Medical, Compassionate, or Extenuating Circumstances](#)

Students missing course work for medical, compassionate, or extenuating circumstances can request academic consideration by completing a request at the [central academic consideration portal](#). Students are permitted one academic consideration request per course per term **without** supporting documentation. Note that supporting documentation is **always** required for academic consideration requests for examinations scheduled by the office of the registrar (e.g., December and April exams) and for practical laboratory and performance tests (typically scheduled during the last week of the term).

Students should also note that the instructor may **designate** one assessment per course per term that requires supporting documentation. This designated assessment is described elsewhere in this document. Academic consideration requests may be denied when flexibility in assessment has already been included. Examples of flexibility in assessment include when there are assessments not required for calculation of the final grade (e.g. 8 out of 10 quizzes) or there is flexibility in the submission timeframe (e.g. 72 hour no late penalty period).

Please note that any academic considerations granted in this course will be determined by the instructor of this course, in consultation with the academic advisors in your Faculty of Registration, in accordance with information presented in this course syllabus. Supporting documentation for academic considerations for absences due to illness should use the Student Medical Certificate or, where that is not possible, equivalent documentation by a health care practitioner.

Policy: [Academic Consideration – Undergraduate Students in First Entry Programs](#)

Procedures: [Student Medical Certificate](#)

Religious Holidays

Students should review the policy for Accommodation for Religious Holidays (Appendix 1). Where a student will be unable to write examinations and term tests due to a conflicting religious holiday, they should inform their instructors as soon as possible but not later than two weeks prior to writing the examination/term test. In the case of conflict with a midterm test, students should inform their instructor as soon as possible but not later than one week prior to the midterm.

Policy: [Accommodation for Religious Holidays](#)

Special Examinations

A Special Examination is any examination other than the regular examination, and it may be offered only with the permission of the Dean of the Faculty in which the student is registered, in consultation with the instructor and Department Chair. Permission to write a Special Examination may be given on the basis of compassionate or medical grounds with appropriate supporting documents. To provide an opportunity for students to recover from the circumstances resulting in a Special Examination, the University has implemented Special Examinations dates.

Policy: [Definitions of Types of Examinations](#)

B. Academic Appeals and Scholastic Offenses

Students can file a **request for relief from academic decisions** if the request is based on one or more grounds listed in the policy. Requests for relief generally fall into three categories, which are also listed in the policy. All requests for relief must be supported by evidence. A request for relief from academic decisions process was formally referred to as an appeal. Refer to the policy and procedures about further details and timelines.

Policy: [Requests for Relief from Academic Decisions](#)

Procedures: [Undergraduate Student Academic Requests for Relief](#)

Scholastic offences are taken seriously, and students are directed to read the appropriate policy, specifically, the definition of what constitutes a scholastic offence.

Policy: [Scholastic Offences](#)

Procedures: [Undergraduate Scholastic Offences](#)

Students may **appeal** some academic and scholastic disciplinary decisions by a Dean or their designate, to the Senate Review Board Academic (SRBA).

Policy: [Senate Review Board Academic Appeals](#)

Procedures: [Senate Review Board Academic Appeals](#)

C. Accessibility Statement

Please contact the course instructor if you require material in an alternate format or if you require any other arrangements to make this course more accessible to you. You may also wish to contact Accessible Education (AE) at 661-2111 x 82147 for any specific question regarding an accommodation.

Policy: [Academic Accommodation for Students with Disabilities](#)

D. Correspondence Statement

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.

E. Discovery Credit Statement

Students are permitted to designate up to 1.0 Discovery Credit course (or equivalent) for pass/fail grading that can be counted toward the overall course credits required for their degree program.

Policy: [Undergraduate Course Credit](#)

Procedures: [Discovery Credits](#)

F. Statement on the Use of Electronic Devices

Assignments and exams in this course will rely extensively on students' ability to problem solve with the assistance of genomic databases containing information related to human genetic disease. As such access to laptops and tablets with internet access is essential during all circumstances.

G. Statement on the Use of Generative Artificial Intelligence (AI)

Related to the above statement about access to electronic devices and the internet, this implies that students will have ready access to be able to query AI chatbots for answers during exams. Students should be aware that the assessments being presented are not reliably able to be answered using AI platforms. Of particular relevance, disputes over acceptability of answers will require documentation of legitimate sources of genetic information, something that AI chatbots routinely hallucinate over. Students are warned here that AI assistance is, in the context of this course, a trap!

H. Turnitin and other similarity review software

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 Western University and [Turnitin.com](https://www.turnitin.com).

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

Policy: [Evaluation of Academic Performance](#)

15. BMSUE Academic Policies and Statements

A. Cell Phone and Electronic Device Policy (for in-person tests and exams)

The Schulich School of Medicine & Dentistry is committed to ensuring that testing and evaluation are undertaken fairly across all our departments and programs. For all tests and exams, it is the policy of the School that any electronic devices, e.g., cell phones, tablets, cameras, smart glasses, smart watch or iPod are strictly prohibited. These devices **MUST** be left either at home or with the student's bag/jacket at the front of the room and **MUST NOT** be at the test/exam desk or in the individual's pocket. Any student found with one of these prohibited devices will receive a grade of zero on the test or exam and this will be documented as a Scholastic Offence. Non-programmable calculators are only allowed when indicated by the instructor. The program is not responsible for stolen/lost or broken devices.

B. Copyright and Audio/Video Recording Statement

Course materials produced by faculty are copyrighted and to reproduce this material for any purposes other than your own educational use contravenes Canadian Copyright Laws. You must always ask permission to record another individual and you should never share or distribute recordings.

C. Rounding of Marks Statement

Across the Basic Medical Sciences Undergraduate Education programs, we strive to maintain high standards that reflect the effort that both students and faculty put into the teaching and learning experience during this course. All students will be treated equally and evaluated based only on their actual achievement. **Final grades** in this course are rounded to the nearest whole number based on the first decimal place. For example, a grade of 74.49 or lower will be rounded to 74, whereas 74.50 or higher will be rounded to 75.

Marks WILL NOT be arbitrarily increased to the next grade or GPA, e.g., a 79 will NOT be increased to an 80, and 84 WILL NOT be increased to an 85, etc. The mark attained is the mark you achieved, and the mark assigned; requests for arbitrary mark increasing will be denied. Marks will be assigned based on assessments in the syllabus and no extra work or tasks will be assigned to increase a mark.

Course grade rounding provisions, as described above, differ from cumulative and term averages. Cumulative and term averages will be calculated to two decimal places and rounded to the nearest whole number with .45 rounded up, for the purposes of admission to and progression in modules, scholarship retention, and Dean's Honour List.

Policy: [Marks/Grades; Definitions of Grades; Grading Scale for Undergraduate Students](#)

16. Support Services

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

Statement on Gender-Based and Sexual Violence

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 the following website:

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.

Other important links:

- [Academic Advising \(Science and Basic Medical Sciences\)](#)
- [Learning Development and Success](#)
- [Office of the Registrar](#)
- [Wellness & Wellbeing](#)
- [Western USC Services](#)

Appendix 1: Western University Academic Policies and Procedures

The policies and procedures listed in this syllabus are outlined in the table below. In some cases, a policy does not include an accompanied procedures document.

Academic Policy	Name of Policy/Procedure	Links
General Policy	Marks/Grades; Definitions of Grades; Grading Scale for Undergraduate Students	Policy
General Policy	Structure of the Academic Year	Policy
Registration, Progression, Graduation	Course Numbering Policy, Essay Courses, and Hours of Instruction	Policy
Registration, Progression, Graduation	Undergraduate Course Credit	Policy • Procedures
Examinations	Definitions of Types of Examinations	Policy
Examinations	Evaluation of Academic Performance	Policy
Examinations	Examination Conflicts	Policy
Rights and Responsibilities	Academic Accommodation for Students with Disabilities	Policy

Rights and Responsibilities	Accommodation for Religious Holidays	Policy
Rights and Responsibilities	Policy on Academic Consideration – Undergraduate Students in First Entry Programs	Policy • Procedures
Rights and Responsibilities	Requests for Relief from Academic Decisions (Undergraduate)	Policy • Procedures
Rights and Responsibilities	Requests for Relief from Academic Decisions (Graduate)	Policy • Procedures
Rights and Responsibilities	Scholastic Offences (Undergraduate)	Policy • Procedures
Rights and Responsibilities	Senate Review Board Academic Appeals	Policy • Procedures