

**Course Number MDSC 541/641
Advanced Genetics**

Instructors:

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Office Hours/Policy on Answering Student Emails

All course communications must occur through your @ucalgary email, and we will respond to emails sent via student's @ucalgary emails within 24 hours M-F.

Time and Location:

Tuesday and Thursday 9:30-10:45. For location see D2L

Prerequisite/Co-Requisite:

Medical Science 341 or Biology 311.

Course Description:

Historical papers will illustrate the foundations of the fundamental principles of modern transmission genetics. Topics including the chromosomal theory of inheritance, the role of pairing and recombination for chromosomal disjunction during meiosis, cytogenetics, the nature of dominant mutations, genetic screens and genetics analysis of developmental pathways. Material covered is drawn from model organisms and humans.

Overarching Theme

This course will focus on milestone events in 21st century genetics that have made the idea of precision human health a reality. Incorporating the underpinning work which first built the human reference genome and adding to it beyond the 'single person genome', we will consider the complex introgression of ancient humans and their genetic legacy in the modern human genome. A better understanding of genetic diversity will allow us to identify, assign and alter the genome in the precision medicine era. We will discuss examples in the identification of rare disease, and developments allowing genome editing. We will introduce the fundamental aspects of epigenetic reprogramming in mammalian model systems and follow how these mechanisms have laid the foundation for iPSC reprogramming, a gateway technology for disease identification, drug screening, and gene-editing-based cures. Using iPSC technology, we will follow the decades of learning that led to the generation of male and female germ cells in vitro and their use in supporting innovative methods for the treatment of human infertility. Throughout will we consider the optic of ethical implications and health inequity issues.

Global Objectives

- To understand modern molecular genetics including discussing different genomes, sequencing, rare disease genetics and, genome editing.

- To have a detailed understanding of epigenetic regulation of cell commitment from totipotency to pluripotency to create an organism.
- To appreciate the application of genetics in stem cell technologies to find solutions for chronic illnesses and reproductive challenges.

Course Learning Outcomes

By the end of this course, students will be able to:

- Understand current genomic technologies, fundamental to modern genetics
- Understand the construction and evolutionary composition of the human genome
- Appreciation the need for the more inclusive pan-genome with impact on precision medicine
- Understand genetic underpinnings of human disease
- Have a fundamental understanding of the epigenome and its remodeling
- Integrated comprehension of the genetic, epigenetic and reproductive implications in advanced treatment of human infertility
- Gain an appreciation of the ethical implications of the genome and its impact for individuals in health and illness

Transferable Skill Development:

Many of the skills and abilities that you are developing in your coursework are transferable to the workforce, graduate and professional studies and other facets of life. Employers seek applicants with transferable skills because they can be an asset in the workplace, regardless of industry or sector. Transferable skills are core skills for your success in building your future career.

The work that you will do in MDSC 541 will help you build the following transferable skills:

- **Verbal Communication:** Learn and share information by presenting, listening, and interacting with others.
- **Critical Thinking:** Actively and skillfully conceptualize, apply, analyze, synthesize, and/or evaluate information (data, facts, observable phenomena, and research findings) to make a reasoned judgement or draw a reasonable conclusion.
- **Information Literacy:** Find, understand, and use information presented through words, symbols, and images
- **Written Communication:** Share ideas and information by using words, images, and symbols.

Learning Resources

No textbook, all required readings will be posted on D2L.

Recommended Textbooks/Readings

No textbook, all required readings will be posted on D2L.

A Note regarding readings

A list of required readings will be outlined on D2L and links and documents will be made available, where possible. Required readings have been chosen carefully to inform you and enhance the lecture material. **Students are REQUIRED to complete assigned readings BEFORE each lecture.** Instructors will proceed in class on the assumption that students have read completely the assigned readings. Students should be aware that many of the readings they will be assigned may be of an unfamiliar nature and style. Students should allot sufficient time to allow for several reads of the assigned material.

Learning Technology Requirements

Brightspace (by D2L) is located on the University of Calgary server and will be used extensively for communication with students. **It is the student's responsibility to ensure that they receive all posted communications and documents and that they receive emails sent by instructors or fellow students through D2L.** Only your @ucalgary.ca email address may be linked to D2L. Please ensure that you are regularly checking your @ucalgary.ca account.

Evaluation

The University policy on grading and related matters is described in section F of the 2023-2024 Calendar.

In determining the overall grade in the course, the following weights will be used:

- 10% Class participation
- 10% Genetics/genomics assignment
- 30% Graphical abstract assignments (5 abstracts)
- 10% Oral graphical abstract presentation
- 10% Oral student-led journal club
- 30% Final assignment

There is no final exam. A student's final grade for the course is the sum of the separate assignments. It is not necessary to pass each assignment separately in order to pass the course.

A Note regarding Writing Assignments:

Writing skills are important to academic study in all disciplines. In keeping with the University of Calgary's emphasis on the importance of academic writing in student assignments (section E.2 of 2023-24 Calendar), writing is emphasized, and the grading thereof in determining a student's mark in this course. The Bachelor of Health Sciences values excellence in writing. Competence in writing entails skills in crafting logical, clear, coherent, non-redundant sentences, paragraphs and broader arguments, as well as skills with the mechanics of writing (grammar, spelling, punctuation). Sources used in research papers must be properly documented. The University of Calgary offers instructional services through the Students' Success Centre's Writing Support Services (<http://www.ucalgary.ca/writingsupport/>) for students seeking feedback on assignments or seeking to improve their general writing skills. Students are **strongly encouraged** to take advantage of these programs.

Grading Scheme:

Letter Grade	Description	Percentage
A+	Outstanding performance	96-100
A	Excellent performance	90-95.99
A-	Approaching excellent performance	85-89
B+	Exceeding good performance	80-84
B	Good performance	75-79
B-	Approaching good performance	70-74
C+	Exceeding satisfactory performance	65-69
C	Satisfactory performance	60-64
C-	Approaching satisfactory performance	57-59
D+	Marginal pass	54-56
D	Minimal pass	50-53
F	Does not meet course requirements	0-49

Missed Components of Term Work:

Students will lose 20% per day late past the deadline for all assignments. In this case, assignments will **NOT** be accepted more than 24 hours after the posted deadline. Students failing to submit any assignment within this time frame will receive a mark of zero. **Students who miss a quiz will receive a mark of zero unless the instructor has been previously notified.** There will be **NO** exceptions to this policy. Please contact the instructors as soon as possible if you have a challenge in meeting a deadline in the course.

As per University Calendar Section G.1.2, **students who are absent from an in-class assessment will receive a mark of zero on the missed component.** Students who are absent are responsible for contacting their instructor to discuss the impact of their missed assessment. Alternative opportunities for completing missed assessments or shifting of the assessment weight **may** be possible but are not guaranteed. Students who are identified as falsifying information related to missed assessments will be subject to investigation for academic misconduct.

Extensions will NOT be granted on any assignment or quizzes in MDSC 541. The only exceptions to this are those in keeping with the University Calendar (debilitating illness, religious conviction, or severe domestic affliction) that are received in writing and with supporting documentation. Traffic jams and late or full buses are common events in Calgary and are **NOT** acceptable reasons for late arrivals to class, meetings and examinations. Please note that while absences are permitted for religious reasons, students are responsible for providing advance notice and adhering to other guidelines on this matter, as outlined in the University Calendar (<https://www.ucalgary.ca/pubs/calendar/current/e-4.html>).

Course Evaluations and Student Feedback

Student feedback will be sought at the end of the course through the Universal Student Rating of Instruction (USRI) and a qualitative student evaluation. Students are welcome to discuss the process and content of the course at any time with the instructor. Students may also address any concerns they may have with Dr. Fabiola Aparicio-Ting, Associate Dean (Undergraduate Health and Science Education) in the Cumming School of Medicine (feapartic@ucalgary.ca).

Attendance

This course is highly interactive, and student attendance at all lectures, and participation in discussions is expected. The genetics/genomics lab can only be attended in person.

Conduct During Lectures

The classroom should be respected as a safe place to share ideas without judgement - a community in which we can all learn from one another. Students are expected to frame their comments and questions to lecturers in respectful and appropriate language, always maintaining sensitivity towards the topic. Students, employees, and academic staff are also expected to demonstrate behaviour in class that promotes and maintains a positive and productive learning environment.

As members of the University community, students, employees and academic staff are expected to demonstrate conduct that is consistent with the University of Calgary Calendar, the Code of Conduct and Non-Academic Misconduct policy and procedures, which can be found at <https://www.ucalgary.ca/student-services/student-conduct/policy> .

Students are expected to take notes during class and should not rely solely on material supplied by the instructors. Instructors may or may not post lectures notes to D2L, at their individual discretion.

Instructors may cover concepts or examples in class that may not be posted to D2L but may be assessed.

Use of Internet and Electronic Communication Devices in Class

The Bachelor of Health Sciences program aims to create a supportive and respectful learning environment for all students. The use of laptop and mobile devices is acceptable when used in a manner appropriate to the course and classroom activities. However, research studies have found that inappropriate/off-topic use of electronic devices in the classroom negatively affects the learning of others during class time.

Students are responsible for being aware of the University's Internet and email use policy, which can be found at <https://www.ucalgary.ca/legal-services/university-policies-procedures/acceptable-use-electronic-resources-and-information-policy>.

UNIVERSITY OF CALGARY POLICIES AND SUPPORTS

Copyright

All students are required to read the University of Calgary policy on Acceptable Use of Material Protected by Copyright (<https://www.ucalgary.ca/legal-services/university-policies-procedures/acceptable-use-material-protected-copyright-policy>) and requirements of the Copyright Act (<https://laws-lois.justice.gc.ca/eng/acts/C-42/index.html>) to ensure they are aware of the consequences of unauthorized sharing of course materials (including instructor notes, electronic versions of textbooks, etc.). Students who use material protected by copyright in violation of this policy may be disciplined under the Non-Academic Misconduct Policy <https://www.ucalgary.ca/pubs/calendar/current/k.html>.

Instructor Intellectual Property

Course materials created by instructors (including course outlines, presentations and posted notes, labs, case studies, assignments and exams) remain the intellectual property of the instructor. These materials may **NOT** be reproduced, redistributed or copied without the explicit consent of the professor. **The posting of course materials to third party websites such as note-sharing sites without permission is prohibited.** Sharing of extracts of these course materials with other students enrolled in the course **at the same time** may be allowed under fair dealing.

Academic Accommodations

It is the student's responsibility to request academic accommodations according to the University policies and procedures listed below. The Student Accommodations policy is available at <https://ucalgary.ca/student-services/access/prospective-students/academic-accommodations>. Students needing an accommodation based on disability or medical concerns should contact Student Accessibility Services (SAS) in accordance with the Procedure for Accommodations for Students with Disabilities (<https://www.ucalgary.ca/legal-services/sites/default/files/teams/1/Policies-Accommodation-for-Students-with-Disabilities-Procedure.pdf>). SAS will process the request and issue letters of accommodations to instructors. For additional information on support services and accommodations for students with disabilities, visit www.ucalgary.ca/access/.

Students who require an accommodation in relation to their coursework based on a protected ground other than disability should communicate this need in writing to Dr. Fabiola Aparicio-Ting (feaparc@ucalgary.ca), Associate Dean (Undergraduate Health and Science Education).

Academic Misconduct

The University of Calgary is committed to the highest standards of academic integrity and honesty. The University of Calgary has created rules to govern all its members regarding the creation of knowledge and the demonstration of knowledge having been learned.

Academic Misconduct refers to student behaviour that compromises proper assessment of a student's academic activities and includes (but is not limited to): cheating, fabrication, falsification, plagiarism, unauthorized assistance, failure to comply with an instructor's expectations regarding conduct required of students completing academic assessments in their courses, and failure to comply with exam regulations applied by the Registrar. **It also includes using of third party websites/services to access past/current course material, essay/assignment writing services, or real-time assistance in completing assessments, seeking answers to assessment questions and similar, whether paid, bartered or unpaid.**

For information of the Student Academic Misconduct Policy and Procedures, please visit; <https://www.ucalgary.ca/legal-services/university-policies-procedures/student-academic-misconduct-policy>.

Additional information is available on the Academic Integrity website at: <https://ucalgary.ca/student-services/student-success/learning/academic-integrity>.

Recording of Lectures

Audio or video recording of lectures (or similar) is prohibited except where explicit permission has been received from the instructor.

Freedom of Information and Protection of Privacy Act

Student information will be collected in accordance with typical (or usual) classroom practice. Students' assignments will be accessible only by the authorized course faculty. Private information related to the individual student is treated with the utmost regard by the faculty at the University of Calgary

Appeals

If there is a concern with the course, academic matter or a grade, first communicate with the instructor. If these concerns cannot be resolved, students can proceed with an academic appeal, as per Section I of the University Calendar. Students must follow the official reappraisal/appeal process and may contact the Student Ombuds' Office (<http://www.ucalgary.ca/student-services/ombuds>) for assistance with this and with any other academic concerns, including academic and non-academic misconduct. Students should be aware that concerns about graded term work may only be initiated **within 10 business days** of first being notified of the grade. <https://www.ucalgary.ca/pubs/calendar/current/i-2.html>

Media Recording

Audio or video recording of lectures (or similar) is prohibited except where explicit permission has been received from the instructor.

Sexual and Gender-Based Violence Policy

The University recognizes that all members of the University Community should be able to learn, work, teach and live in an environment where they are free from harassment, discrimination, and violence. The University of Calgary's sexual violence policy guides us in how we respond to incidents of sexual violence, including supports available to those who have experienced or witnessed sexual violence, or those who are alleged to have committed sexual violence. It provides clear response procedures and timelines, defines complex concepts, and addresses incidents that occur off-campus in certain

circumstances. Please see the policy available at <https://www.ucalgary.ca/legal-services/sites/default/files/teams/1/Policies-Sexual-and-Gender-Based-Violence-Policy.pdf>.

Resources for Support of Student Learning, Success, Safety and Wellness

Student Success Centre <http://www.ucalgary.ca/ssc/>
Student Wellness Centre <http://www.ucalgary.ca/wellnesscentre/>
Student Advocacy and Wellness Hub (CSM)
<https://cumming.ucalgary.ca/mdprogram/current-students/student-advising-wellness>
Distress Centre <http://www.distresscentre.com/>
Library Resources <http://library.ucalgary.ca>

Wellness and Mental Health Resources

The University of Calgary recognizes the pivotal role that student mental health plays in physical health, social connectedness and academic success, and aspires to create a caring and supportive campus community where individuals can freely talk about mental health and receive supports when needed. We encourage you to explore the excellent mental health resources available throughout the university community, such as counselling, self-help resources, peer support or skills-building available through the SU Wellness Centre (<https://www.ucalgary.ca/wellnesscentre/services/mental-health-services>) and the Campus Mental Health Strategy (<http://www.ucalgary.ca/mentalhealth/>).

Student Ombuds' Office

The Student Ombuds' Office supports and provides a safe, neutral space for students. For more information, please visit www.ucalgary.ca/student-services/ombuds/ or email ombuds@ucalgary.ca

BHSc Student Faculty Liaison Committee (SFLC)

The BHSc SFLC, with elected representatives from all majors, serves to raise issues of interest to BHSc students to the program administration, including items pertaining to curriculum, scheduling and events. A list of current representatives can be found on the BHSc website.

Student Union (SU) Information

The SU Vice-President Academic can be reached at (403) 220-3911 or suvice@ucalgary.ca; the SU representatives for the Cumming School of Medicine can be reached at medrep1@su.ucalgary.ca or medrep2@su.ucalgary.ca.

Student Success Centre

The Student Success Centre provides services and programs to ensure students can make the most of their time at the University of Calgary. Our advisors, learning support staff, and writing support staff assist students in enhancing their skills and achieving their academic goals. They provide tailored learning support and advising programs, as well as one-on-one services, free of charge to all undergraduate and graduate students. For more information visit: <https://www.ucalgary.ca/student-services/student-success>

Emergency Evacuation/Assembly Points

As part of the University of Calgary Emergency Evacuation plan, students, faculty, and staff should locate the closest Assembly Point in case of Fire Alarm. Safety signage is posted throughout the campus showing the locations and the possible route to these locations. All students, faculty, and staff are expected to promptly make their way to the nearest Assembly Point if the Fire Alarm is activated. No one is to return into campus facilities until an all clear is given to the warden in charge of the Assembly

Area. For more information, see <https://www.ucalgary.ca/emergencyplan/building-evacuation/assembly-points>

Safewalk

Campus security will escort individuals, day or night, anywhere on campus (including McMahon Stadium, Health Sciences Centre, Student Family Housing, the Alberta Children's Hospital and the University LRT station). Call 403-220-5333 or visit <http://www.ucalgary.ca/security/safewalk>. Use any campus phone, emergency phone or the yellow phone located at most parking lot pay booths. Please ensure your personal safety by taking advantage of this service.

Class Schedule

The following is a list of topics for class, associated readings, and assignment due dates. Please note that unforeseen circumstances may cause changes to the schedule with respect to the timing of topics and readings. Students will be notified of all changes in a timely manner by way of email and D2L announcements.

Date	Topic	Assignments
Sept 5	Introduction to the human genome	
Sept 7	Human Genome sequencing	
Sept 12	Genetics of disease/rare disease	Graphical abstract 1
Sept 14	Craniofacial syndromes and genetic modifiers	
Sept 19	Genetics/Genomics in class lab	
Sept 21	Genome editing- the Brave new World, ethics	Genomics
Sept 26	Privacy and permissions	
Sept 28	Ancestral genomes in the modern era	Graphical abstract 2
Oct 3	Peer learning- graphical abstract presentations	
Oct 5	Structured paper discussion on ancient DNA	
Oct 10	Ancient genomes in us	Student-led journal club
Oct 12	Pangenome	Student-led journal club
Oct 17	The role of the epigenome	Graphical abstract 3
Oct 19	Transgenerational epigenetic inheritance	
Oct 24	The thrift hypothesis-Barker hypothesis	Graphical abstract 4
Oct 26	Precision health implications	
Oct 31	Leveraging the genome for reproductive health	
Nov 2	Epigenetic reprogramming- germ cells	Graphical abstract 5
Nov 7	Building tools for understanding development- ES and iPSC	
Nov 14	Reading break	
Nov 16	Reading break	
Nov 23	Experimental reprogramming- turning back time- DNA methylation clock	Draft of final paper concept
Nov 28	Paper discussion	Student-led journal club
Nov 30	Rejuvenation and the ethics of editing in human reproduction	
Dec 5	Discussion day	