

MDSC 341
Principles of Human Genetics

Instructors:

Course coordinators and instructors:

Dr. Jillian Parboosingh jillian.parboosingh@albertaprecisionlabs.ca

Dr. Amanda Tyndall amanda.tyndall@albertaprecisionlabs.ca

Instructors:

Dr. Sarah Barclay sarah.barclay@albertaprecisionlabs.ca

Dr. Eliza Phillips eliza.phillips@albertahealthservices.ca

Office Hours/Policy on Answering Student Emails

Meetings with the instructor are available after lecture or by appointment (by Zoom, phone or in person). Teaching assistants are not available outside of scheduled tutorial time. All questions regarding course material should be directed to the instructor.

Students may contact the course instructor by email. Please type MDSC 341 as the subject. The instructor will endeavour to respond within 2 working days.

Teaching Assistant:

Harmony Fong harmony.fong@ucalgary.ca

Emma Gillesse emma.gillesse@ucalgary.ca

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Time and Location:

LECTURES: Sept. 3, 5, 10, 12, 17, 19, 24, 26, Oct. 1, 3, 8, 10, 15, 17, 24, 29, 31, Nov. 5, 7, 19, 21, 26, 28, Dec. 3, 5

Days/Time: Tuesdays, Thursdays 3:00-4:20 PM

Location: See D2L for location details

The instructors will provide lecture material in either pdf or PowerPoint form on D2L. However, there is no guarantee that it will be posted prior to the lecture. During lectures, material posted will be expanded upon and discussed. Therefore, students should not rely on the web material to obtain all the material taught in a class.

There will be no lecture on October 22 (replaced by Midterm Exam, 3:00 – 5:50 pm).

Fall Term Break is November 10-16; there are no lectures November 12 and 14.

TUTORIALS:

Days/Time: Tuesdays, 4:30 – 5:50 pm: Sept. 3, 10, 17, 24, Oct. 1, 8, 15, 29, Nov. 5, 19, 26, Dec. 3

Thursdays, 4:30 – 5:50 pm: Sept. 5, 12, 19, 26, Oct. 3, 10, 17, 31, Nov. 7, 21, 28, Dec. 5

Location: See D2L for location details

There will be no tutorials October 22 or 24 (Midterm Exam, October 22, 3:00 – 5:50 pm).
Fall Term Break is November 10-16; there are no tutorials November 12 and 14.

Prerequisite/Co-Requisite:

Biology 241, 243 and admission to the BHSc Honours program.

Course Description:

Introduction to principles in human genetics including Mendelian and chromosomal basis of inheritance, chromosomal abnormalities, pedigree analysis, mutations, and molecular, metabolic, population and clinical genetics. Studies of model organisms and genomics will be included as required. Incorporates problem-based learning to establish analytical skills in genetics.

Overarching Theme

Genetics is the study of biological information, how it is stored, replicated, used and passed on to the next generation. Genetics plays an ever-increasingly important role in medicine and our world today. It is now recognized that genetics is important not only in rare monogenic disorders but in common disorders such as cancer, diabetes and cardiovascular disease (multifactorial diseases), in the way we react to drugs, other therapies and the environment (pharmacogenomics, envirogenetics) and in the aging process.

The purpose of this course is to provide the student with a foundation in genetics, including Mendelian and chromosomal basis of inheritance, chromosomal abnormalities, pedigree analysis, mutations, and molecular, metabolic, population and clinical genetics. Studies of model organisms and genomics will be included as required. Incorporates problem-based learning to establish analytical skills in genetics.

Global Objectives

- Promote active learning, critical thinking and an inquiry approach to genetics
- Provide students with opportunities to develop and practice problem-solving skills
- Integrate Mendelian principles, molecular biology, and human genetics

Course Learning Outcomes

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

- Utilize genetic terminology correctly
- Explain the underlying basis of inheritance
- Illustrate how DNA provides the genetic blueprint for cellular structures and processes
- Predict the implications of genetic variation
- Discuss how genes contribute to health and disease
- Solve a variety of genetic problems

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 341 will help you build the following transferable skills:

- **Collaboration:** Work respectfully with others from different backgrounds, cultures, and countries.
- **Verbal Communication:** Learn and share information by presenting, listening, and interacting with others.
- **Creativity and Innovation:** Find different and better ways to do things, being curious, thinking imaginatively.
- **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.
- **Digital Skills:** Use digital technologies like computers, social media, virtual meeting platforms, and the internet.
- **Information Literacy:** Find, understand, and use information presented through words, symbols, and images
- **Numeracy:** Use mathematical information such as numbers, symbols, words, and graphics to do tasks.
- **Problem solving:** Identify an issue, find and implement a solution, and assess whether the situation has improved.
- **Written Communication:** Share ideas and information by using words, images, and symbols.

Learning Resources

Required Textbook: Michael L. Goldberg, Janice Fischer. Genetics: from genes to genomes 8th edition.

Either the online access or hardcopy are acceptable. *The 7th edition is a reasonable alternative and used copies may be available. However, students who choose to use the 7th edition will be responsible for identifying the corresponding chapters and page numbers for suggested readings and problems.*

Online Access: estimated cost is \$70 for 180-day rental ebook. You may obtain a code at the bookstore or buy directly from McGraw Hill at this link (see D2L for instructions):

<https://www.mheducation.ca/higher-education>

Hardcopy: estimated cost is \$150, available at the Medical School Bookstore.

A Note regarding readings

Some classes will have required activities (readings, videos, etc) to be completed **BEFORE** lecture. For other lectures, students are encouraged to complete assigned readings and activities before each lecture. These will be clearly posted on D2L and additional links and documents will be made available. Suggested readings and activities have been chosen to inform you and support the lecture material.

Instructors will proceed in class on the assumption that students have completed the pre-assigned activities. Students should be aware that the content may be of an unfamiliar nature and style. **Students should allot sufficient time to allow for completion 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 2024-2025 Calendar.

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

Online quizzes (10%)

Each week prior to tutorial, students will be given access on D2L to a brief online quiz about the tutorial topics. The quiz must be completed by 11:59 pm the Monday before tutorial (exception tutorial 1 which will be available all week). Students will be allowed two attempts of the quiz with the highest score being awarded.

Tutorials (10%)

The tutorials provide an opportunity for students to work through problems and learn essential concepts in genetics in small groups with the assistance of a teaching assistant. Each week prior to tutorial, students will be given access on D2L to the tutorial problem set. During the tutorial, the teaching assistants will lead the students through the problems and additional exercises. The tutorials are highly interactive, and participation is required. **Students must print and read the tutorial problems prior to class and bring their copy with them.** Students are not expected to complete the problems prior to tutorial. One additional question will be given during the final 15-20 minutes of the tutorial. This is an open book question, in which students may use their class notes, work in groups, and may ask their TA questions. This question is to be completed by each student (whether or not they work in groups) and is to be handed in to the TA at the end of tutorial. Please note that, other than calculators, no electronics other than tablets may be used in tutorial without permission of the Student Accessibility Services or TA.

There are 12 tutorials. The bottom two grades or up to two missed tutorials will not count towards the tutorial mark. Students missing tutorials, are encouraged to submit tutorial questions the following week, not for marks but to assist with assessing their understanding and obtaining the correct answers for study purposes.

Assignments (20%)

Four assignments will be posted on D2L that will be based on the concepts in class and tutorial.

Problem set	Assigned	Due (start of class)	Weight
Chromosomes, Gene Expression and regulation	Sep 17	Sep 24	5%
Mendelian genetics, Mapping and Mutation	Oct 1	Oct 8	5%
Genetic Diagnostics	Nov 7	Nov 19	5%
Cell Cycle and Cancer	Nov 28	Dec 5	5%

Completed assignments are to be handed in at the beginning of class on the due date indicated. Assignments will be assessed by the TAs. Marks will be given for both the answer and work. All work shown must support the answer provided. Late assignments will not be accepted, and illegible assignments will not be marked.

Exams (60%)

Exams will have short answer and problem-solving questions and will be similar to the tutorial questions, assignments and recommended questions from the text. Part marks will be given for work shown. The midterm must be written in pen. Students may bring a simple, non-programmable calculator. No other electronic devices (including smart watches and phones) are permitted.

Midterm: October 22, 3-5:50 PM, Theatre One and Clara Christie (students will be assigned to one of the rooms)

Material, assignments and readings up to and including October 10, and Tutorials 1-7

Weight: 30%

Final Exam: 3 h, date during the final exam period, scheduled by the registrar

Material, assignments, and readings and tutorials after October 10, including tutorials 8 – 12.

Weight: 30%

Note: A student's final grade for the course is the sum of the separate evaluations. It is not necessary to pass each evaluation 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 2024-25 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 to complete each assessment must be properly documented, unless otherwise noted by the instructor. 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:

At each instructor's discretion, late assignments will not be accepted and will automatically receive a mark of zero.

As per University Calendar Section G.1.2, students who are absent from tutorial 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 341. 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 new UCalgary Course Experience Survey 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

Regular attendance is advised in order to succeed in MDSC 341. 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. In-class discussion and all content presented in class, including concepts and examples, can constitute substantial learning and can be considered for assessment.

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://ucalgary.ca/student-services/student-conduct/policy> .

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>.

Use of Artificial Intelligence Tools

Generative Artificial Intelligence (AI), and specifically foundational models that can create writing, computer code, and /or images using minimal human prompting includes not only GPT-4 (and its siblings ChatGPT and Bing), but many writing assistants that are built on this or similar AI technologies.

The use of generative artificial intelligence tools and apps **is strictly prohibited** in all course assignments unless explicitly stated otherwise by the instructor in this course. This includes ChatGPT and other AI writing and coding assistants. Use of generative AI in this course may be considered use of an unauthorized aid, which is a form of cheating and a breach of academic integrity subject to Academic Misconduct procedures. You could be asked to provide evidence of your own work.

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/legal-services/university-policies-procedures/student-non-academic-misconduct-policy>

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>).

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) by students 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://calendar.ucalgary.ca/pages/e31a7115dca740ec83579e946d4a4193>

Recording of Lectures

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/university-policies-procedures/sexual-and-gender-based-violence-policy>.

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.