

Medical Sciences 543 (MDSC 543)
Advanced Molecular Genetics

Instructors:

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Office Hours/Policy on Answering Student Emails

Meetings with the instructors are available after lecture or by appointment. All questions regarding course material should be directed to the appropriate instructor.

Students may contact course instructors by email. Please type MDSC 543 as the subject. Instructors will endeavour to respond within 2 working days.

Time and Location:

Days/Time: Wednesdays 2 – 4:50 PM

Location: At a minimum, January 12, 19, and 26, classes will be held over Zoom; links will be distributed via D2L. Thereafter, dependent upon direction from the University of Calgary and Alberta Health Services, classes may resume in-person and will be located in HSC G801E, unless notified otherwise.

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.*

Prerequisite/Co-Requisite:

Prerequisite - MDSC 341 or BIOL 311

Course Description:

This is the third advanced genetics course which focuses on the molecular genetic basis of human diseases. Building on fundamental genetic principles, a teaching team of laboratory geneticists will

introduce students to the types of genetic changes and mutational mechanisms associated with human diseases. A major focus of the course is on molecular genetic pathology and how pathogenicity is determined.

Overarching Theme

Advanced Molecular Genetics - Genes to Genomes and Understanding Human Genetic Diseases. The focus will be on applied molecular genetic analysis with an emphasis on human health and disease. Each class will consist of a seminar presentation by the instructor and an interactive activity in which students will participate. This course is double listed as a senior graduate course (MDSC 543) and graduate course (MDSC 641.03).

Global Objectives

The goals of the course are to:

- ◆ 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 have the necessary knowledge and skills to be able to:

1. Recognize the complexity of the human genome.
2. Describe the genetic mechanisms underlying the etiology of human disorders /conditions.
3. Explain the use of molecular genetic techniques for the identification of human genetic variation.

Learning Resources

None required.

Recommended Textbooks/Readings

Human Molecular Genetics, 5th Edition - Strachan and Read (2018)

Thompson and Thompson Genetics in Medicine, 8th Edition – Nussbaum, McInnes and Willard (2016)

Genetics and Genomics in Medicine, Strachan, (2014)

A Note regarding readings

A list of required readings for all course sections 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 2021-2022 Calendar.

Students are expected to write effectively to communicate their ideas and responses to questions. While writing skills are not directly being evaluated, the ability to write legibly and communicate answers (including use of the genetic terms introduced in class) impacts evaluation. Illegible answers will not be marked; this includes very small handwriting or poorly scanned documents.

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

Assignments (40%)

Assignment	Description	Weight %	Due Date and Time
#1	The Human Genome and its Complexity/Gene Expression and Regulation problem set	10	Jan. 26, 2022, 2 pm
#2	Molecular Genetic Pathology problem set	10	Feb. 9, 2022, 2 pm
#3	Cell-free DNA/mitochondrial genetics problem set	10	Mar. 2, 2022, 2 pm
#4	Structured Paper review	10	Mar. 16, 2022, 2 pm

In-class participation (10%)

Participation in class discussions

In-class written evaluation (25%)

Short answer format, Mar. 23, 2022.

Presentation to class (25 %)

Presentation on approved topic to class, Mar. 30 or Apr. 6, 2022

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 2021-22 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
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	Did not meet course requirements	0-49

Missed Components of Term Work:

Late assignments will not be accepted and will automatically receive a mark of zero. Students who miss a quiz or in class assignment will receive a mark of zero unless the instructor has been previously notified. There will be NO exceptions to this policy.

It is the agreement of all Faculty involved in MDSC 543 that **extensions will NOT be granted** on any assignment or quizzes. 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. Ebba Kurz, Associate Dean (Undergraduate Health and Science Education) in the Cumming School of Medicine (kurz@ucalgary.ca).

Attendance

Attendance at all lectures is expected of all students.

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/legal-services/university-policies-procedures>.

Students are expected to take notes during class and should not rely solely on material supplied by the instructors.

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/policies/files/policies/electronic-communications-policy.pdf>.

UNIVERSITY OF CALGARY POLICIES AND SUPPORTS

Copyright

All students are required to reach the University of Calgary policy on Acceptable Use of Material Protected by Copyright (<https://www.ucalgary.ca/policies/files/policies/acceptable-use-of-material-protected-by-copyright-policy.pdf>) 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/policies/files/policies/procedure-for-accommodations-for-students-with-disabilities.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. Ebba Kurz, Associate Dean (Undergraduate Health and Science Education) (kurz@ucalgary.ca).

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://ucalgary.ca/policies/files/policies/student-academic-misconduct-policy.pdf>
<https://ucalgary.ca/policies/files/policies/student-academic-misconduct-procedure.pdf>

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 or it is an approved accommodation.

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/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>

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 and gender-based violence policy guides us in how we respond to incidents of sexual and gender-based violence, including supports available to those who have experienced or witnessed sexual or gender-based violence, or those who are alleged to have committed sexual or gender-based 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/
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/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 suvpaca@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, and assignment due dates. Please note that unforeseen circumstances may cause changes to the schedule with respect to the timing of topics. Students will be notified of all changes in a timely manner by way of email and D2L announcements. The in-class evaluation date is firm and will not be altered.

Date	Module / Topics	Instructor/Guest Lecturer	Assignments Due Dates
Jan. 12, 2022	Human Genome and its complexity	Dr. Argiropoulos	
Jan. 19, 2022	Gene expression and regulation	Dr. Racacho	
Jan. 26, 2022	Molecular genetic pathology I: SNVs, indels, dynamic expansions	Drs. Lamont, Parboosingh	Assignment 1
Feb. 2, 2022	Molecular genetic pathology II: CNVs, SVs, pseudogenes	Drs. Lamont, Parboosingh	
Feb. 9, 2022	Cell free DNA: non-invasive prenatal testing, circulating tumour DNA	Dr. Tyndall (guest lecturer)	Assignment 2
Feb. 16, 2022	Mitochondrial Genetics	Dr. Shutt (guest lecturer)	
Feb. 20 – 26, 2022 Term Break			
Mar. 2, 2022	Cancer Genetics	Dr. Box (guest lecturer)	Assignment 3
Mar. 9, 2022	Clinical NGS-based tests WES analysis	Drs. Lamont, Racacho	
Mar. 16, 2022	Pharmacogenomics	Dr. Bousman (guest lecturer)	Assignment 4
Mar. 23, 2022	Final in-class written assessment	Dr. Parboosingh	
Mar. 30, 2022	Student presentations	Drs. Argiropoulos, Lamont, Parboosingh, Racacho	
Apr. 6, 2022	Student presentations	Drs. Argiropoulos, Lamont, Parboosingh, Racacho	