The University of Calgary
Bachelor of Health Sciences
Cumming School of Medicine

Academic Year 2023-2024

MDSC 545
Genomics

Instructors:

Dr. Lemuel Racacho (lemuel.racacho@ucalgary.ca, 403-955-5385)
Dr. Tamas Revay (tamas.revay@ucalgary.ca, 403-955-7377)

Office Hours/Policy on Answering Student Emails

Please book appointments by email. Email communications with the instructor and the TA are welcome. All efforts will be made to respond to emails within 48 hours. Substantive issues are to be addressed in-person, either in class or during an appointment.

Teaching Assistant:

Eva Zaffarini (eva.zaffarini@ucalgary.ca)

Time and Location:

Fall 2023, Tuesdays and Thursdays, 1:30 pm to 2:50 pm
For location see D2L

Prerequisite/Co-Requisite:

Medical Science 408 or Cellular, Molecular and Microbial Biology 413 or consent of the instructor.

Course Description:

Examine the strategies and techniques, including massively parallel sequencing, used in genomic and genetic studies. Review how model systems and genome editing are used to establish the functional consequences of genomic variation. Students can expect to gain a comprehensive understanding and broad appreciation of the significance of genomic information in context of rare and common human diseases, and genome biology.

Overarching Theme

Students will be provided foundations to understand the structural and functional elements of the human genome. Moreover, examples of genetic studies and the use of genomic technologies, such as next-generation sequencing, will be presented to highlight an up-to-date insight into the structure of the human genome. In addition, lectures on hot-topic research areas within genomics, such as cytogenomics, cancer genomics, rare disease genomics, population, evolutionary genomics, ethics etc. will serve to deepen the understanding of these foundations.

Global Objectives

To inspire and spark interest in genomics in an interactive classroom environment that is engaging and intellectually challenging.

Course Learning Outcomes

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

1. Recognize how genomic information has been used to identify the underlying cause of genetic disorders and enhance our understanding of biological systems.
2. Be familiar with current genomic methodologies and databases and develop an appreciation for the quantitative and qualitative concepts underlying these processes.
3. Appreciate the complex ethical, legal and social issues that accompany genomic data and its applicability.

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 **MDSC545** 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.
- **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
- **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/ Recommended Textbooks/Readings

There is no assigned textbook for this course. Required reading from primary literature will be assigned. Videos, blogs and glossary of terms will be provided to emphasize key concepts through a variety of media.

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 all assigned readings.*** *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:

Assessment Components: The University policy on grading-related matters is outlined in the Academic Calendar.			
Assessment Methods	**Description**	**Weight %**	**Due Date and Time (in MST)**
Assignments	Two in-class quiz and one take home assignment based on the foundation lectures	3x10%	Sept. 14, Sept. 26, Oct. 10
Term paper	Written assignment on assigned topic relating to Genomics	20%	Nov. 21
Oral Presentation	Presentation on assigned topic relating to Genomics.	40%	TBA, Nov. 21-Dec. 5
Participation	Classroom or zoom participation	10%	

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.

Guidelines for submitting assignments:

The two in-class quizzes are closed book, intended to assess the knowledge acquired from the foundational lectures and discussions. Quiz #1: lectures 1-3, quiz #2: lectures 4-6. The assignments will be scheduled at the beginning of given class for max 20 minutes and will consist of a mix of multiple choice, fill-in-the-blank, short answer (1-2 sentences) type questions.

The third assignment is a take-home question set. The written short answers require literature reading and data-mining. The topics will be defined in class.

Oral presentation: The literature on human genomics is extensive and diverse. During the lecture portion of this course it will not be possible to cover all the topics in this field. Therefore, you are required to review a topic. A list of topics is provided. To make it interesting, you are asked to pair with one of your peers to discuss the two contrasting sides/aspects of the given question. It is not a formal debate and the assessment does not aim to select a winner. Each member of the pair should present the chosen aspect of the topic with clarity, including rigorous scientific background and personal perspective. Both members of the pair should create a ~15 min. oral presentation, supported by ppt slides. Be creative. After both presentations the class opens for discussion of the ideas, sides, and opinions. Participation in this discussion is part of the evaluation, responses for questions, asking questions from the other team member is encouraged.
A list of topics is also provided to be discussed alone, if pairs cannot be formed. The presenter addresses the topic to the class in a 15 min oral presentation and take part of the following discussion.

The term paper is a summary of the oral presentation on the decided topic. It is a max 2 pages long formal written document. The structure of the document should include an Introduction, Discussion, Conclusion and References. It is preferred to create the written summary first, given this will incorporate all researched background and personal perspective that is than easily shaped into the oral presentation. Please use Times New Roman, font 12, line spacing 1 or 1.5 and Justified. The document will be submitted electronically through D2L in a designated dropbox by the due date at 1:30pm.

FINAL EXAMINATIONS: There is no final exam for this 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:
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 545. 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 (feaparic@ucalgary.ca).

Attendance

As 10% of the final mark is based on participation, we strongly advise that you show up to all teaching sessions (in person or on Zoom), ask questions and engage in class discussions.

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, Associate Dean (Undergraduate Health and Science Education) in the Cumming School of Medicine (feaparic@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://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

Please refer to the following statement on media recording of students: https://elearn.ucalgary.ca/wp-content/uploads/2020/05/Media-Recording-in-Learning-Environments-OSP_FINAL.pdf

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 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, associated readings, and assignment / exam 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. The exam dates are firm and will not be altered.

Date	Topic	Instructor	Assignments Due Dates
Tuesday, September 05, 2023	Introduction, Course overview, historical perspectives	Dr. Tamas Revay	
Thursday, September 07, 2023	The human genome - 1	Dr. Tamas Revay	
Tuesday, September 12, 2023	The human genome - 2	Dr. Lemuel Racacho	
Thursday, September 14, 2023	Genomics technologies - 1	Dr. Lemuel Racacho	in-class quiz #1
Tuesday, September 19, 2023	Genomics technologies - 2	Dr. Tamas Revay	
Thursday, September 21, 2023	Bioinformatics	Dr. Lemuel Racacho	
Tuesday, September 26, 2023	Medical genomics	Dr. Tamas Revay	in-class quiz #2
Thursday, September 28, 2023	Variant analysis	Dr. Ryan Lamont	take-home assignment available
Tuesday, October 03, 2023	Cytogenomics	Dr. Tamas Revay	
Thursday, October 05, 2023	Population Genomics / GWAS	Eva Zaffarini	
Tuesday, October 10, 2023	GenEthics	Amy Davis Natasha Lemiski	take-home assignment due
Thursday, October 12, 2023	Epigenomics	Dr. Lemuel Racacho	
Tuesday, October 17, 2023	Cytogenomic laboratory visit	Dr. Tamas Revay	
Thursday, October 19, 2023	Evolutionary Genomics	Dr. Sam Yeaman	
Tuesday, October 24, 2023	Molecular Genetic lab. visit	Dr. Lemuel Racacho	
Thursday, October 26, 2023	Cancer genomics	Dr. Paola Neri	
Tuesday, October 31, 2023	Metagenomics	Dr. Laura Sycuro	
Thursday, November 02, 2023	Genome editing	Dr. Sarah Childs	
Tuesday, November 07, 2023	Mitochondrial Genomics	Dr. Tim Shutt	
Thursday, November 09, 2023	example presentation/debate	Dr. Tamas Revay, Dr. Lemuel Racacho	written and oral assignment available
Tuesday, November 14, 2023	Term break No class		

Date	Topic	Instructor	Assignments Due Dates
Thursday, November 16, 2023	Term break No class		
Tuesday, November 21, 2023	Student presentation/debate	Student led	written assignment due
Thursday, November 23, 2023	Student presentation/debate	Student led	
Tuesday, November 28, 2023	Student presentation/debate	Student led	
Thursday, November 30, 2023	Student presentation/debate	Student led	
Tuesday, December 05, 2023	Student presentation/debate	Student led	