

## **MDSC 543 / MDSC 641.03**

### **Advanced Genetics II (Advanced Molecular Genetics)**

#### **Instructors:**

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#### **Office Hours/Policy on Answering Student Emails**

For individual concerns or inquiries, emails will be responded to within 48 hours. It is preferred that broader content questions be shared on the D2L course page to permit sharing and for open class discussion.

#### **Time and Location:**

Wednesdays 2 - 4:50 pm  
January 10 – April 11, 2018.

Genetics Laboratory Conference Room\*, ACH B0-170  
Lower Level, Alberta Children's Hospital

\*Please sign in at the front of the laboratory area

#### **Prerequisite/Co-Requisite:**

None other than permission of the instructor

#### **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 the theme of relevance to issues in health and society. 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).

## Learning Objectives

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

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

## Required Textbooks

*None required.*

## Recommended Textbooks/Readings

Human Molecular Genetics, 4th Edition - Strachan and Read (2011)

Thompson and Thompson Genetics in Medicine, 8<sup>th</sup> Edition – Nussbaum, McInnes and Willard (2015)

Genetics and Genomics in Medicine, Strachan, (2014)

*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.*

## Evaluation

The University policy on grading and related matters is described in section F.2 of the 2017-2018 Calendar.

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

40% Take-home assigned questions/exercises

Due Dates: #1 January 31; #2 February 7; #3 March 7; #4 March 14

10% Class participation in discussions – 10% per class (except Feb 21, March 28; April 4 and 11)

25% In-class written evaluation\* – March 28

25% Presentation to class, on selected topic April 4

*\* calculators will be permitted*

*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 2017-18 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). The University of Calgary offers a number of 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:

A+ 97-100%	B+ 80-84%	C+ 65-69%	D+ 54-56%
A 90-96%	B 75-79%	C 60-64%	D 50-53%
A- 85-89%	B- 70-74%	C- 57-59%	F 0-49%

### Missed Components of Term Work:

It is the agreement of all Faculty and Staff involved in MDSC 641.03 that extensions will NOT be granted on any assignment or evaluation. Late assignments will not be accepted and will automatically receive a mark of zero. The only exceptions to this are those in keeping with the University Calendar (illness, religious conviction, or domestic affliction) that are received in writing and with supporting documentation.

### Desire2Learn (D2L)

Desire 2 Learn 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 s/he gets all posted communications and documents and that s/he receives 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.

If you need help accessing or using D2L, please visit the Desire2Learn resource page for students: <http://elearn.ucalgary.ca/d2l-student/>.

### Policies Governing the Course:

#### Attendance

Students are expected to come to class prepared and actively participate in class. As indicated above, 10% of the class grade is reserved for participation.

### **Conduct During Lectures**

Students are expected to conduct themselves in a mature and courteous manner during ALL lectures. Students are expected to frame their comments and questions to lecturers in respectful and appropriate language, always maintaining sensitivity towards the topic. Students are expected to fully participate in class discussions, being respectful of all points of view.

**Students are expected to take notes during each session and should not rely solely on handout material supplied by the instructors.**

### **Electronic Devices**

The Bachelor of Health Sciences program aims to create a supportive and respectful learning environment for all students. Research studies have found that student use of electronic devices (laptops, tablets, etc) in the classroom negatively affects the learning of both the user and those sitting nearby. Inappropriate use of laptops is also disruptive to your fellow classmates and disrespectful to the lecturer. The use of laptops and other electronic note-taking devices is permitted; however, their use in the classroom should be for course-related work/note-taking only. Please do **NOT be to surf the web, check email or do other unrelated work**. Students who use their laptops inappropriately or are otherwise disruptive during lectures will be asked to leave.

Cell phones (or similar devices) should **be turned off** (not merely silent) upon entering the classroom. Sending/receiving text messages or leaving the class to take calls is disruptive to the entire class and will not be tolerated unless absolutely necessary. Students who disregard this rule during lectures or tutorials will be asked to leave. These items are not permitted under any circumstance during exams/quizzes, etc.

### **Copyright**

It is the responsibility of students and professors to ensure that materials they post or distribute to others comply with the Copyright Act and the University's Fair Dealing Guidance for Students ([library.ucalgary.ca/files/library/guidance\\_for\\_students.pdf](http://library.ucalgary.ca/files/library/guidance_for_students.pdf)). Further information for students is available on the Copyright Office web page (<http://library.ucalgary.ca/copyright>)

### **A Note Regarding Instructor Intellectual Property**

Generally speaking, course materials created by professor(s) (including course outlines, presentations and posted notes, labs, case studies, assignments and exams) remain the intellectual property of the professor(s). 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 Based on Disability or Medical Condition**

It is the student's responsibility to register with Student Accessibility Services to be eligible for formal academic accommodation 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\\_0.pdf](https://www.ucalgary.ca/policies/files/policies/procedure-for-accommodations-for-students-with-disabilities_0.pdf)). If you are a student who may require academic accommodation and have not registered with Student Accessibility Services, please contact their office at (403) 220-8237; <http://www.ucalgary.ca/access/>. Students will be provided with all necessary accommodations to ensure equal opportunity to succeed in this course. Please provide the instructor your accommodation letter from Student Accessibility Services within 14 days after the start of this course so that all needed arrangements for exams and assignments can be made.

### **Accommodations on Protected Grounds other than Disability**

Students who require an accommodation in relation to their coursework based on a protected ground other than disability, should communicate this need, preferably in writing, to the designated BHSc program contact, Mrs. Jennifer Logan ([jljlogan@ucalgary](mailto:jljlogan@ucalgary)), or to Dr. Ebba Kurz, Associate Dean, Undergraduate Health and Science Education, Cumming School of Medicine. Students who require an accommodation unrelated to their coursework or the requirements for a graduate degree, based on a protected ground other than disability, should communicate this need, preferably in writing, to the Vice-Provost (Student Experience). For additional information on support services and accommodations for students with disabilities, visit [www.ucalgary.ca/access/](http://www.ucalgary.ca/access/).

### **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. These rules are contained principally in Sections J to L of the *University of Calgary Calendar*. Students are expected to be familiar with these standards and to uphold the policies of the University in this respect. The Calendar also stipulates the penalties for violating these rules. Please know that the University and the Cumming School of Medicine take these rules seriously. **All incidences of academic dishonesty in this course, such as cheating and plagiarism, will be reported to the Associate Dean for investigation;** infractions will be noted on the record of a student found to be guilty.

### **Recording of Lectures**

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

### **Other Important Information**

#### **Freedom of Information and Protection of Privacy Act**

This course is conducted in accordance with the Freedom of Information and Protection of Privacy Act (FOIP); students should identify themselves on written assignments (exams and term work) by their name and ID number on the front page and ID on each subsequent page.

Work assigned to you by your course instructor will remain confidential unless otherwise stated before submission. The assignment cannot be returned to anyone else without your expressed permission to the instructor. Grades will be made available on an individual basis and students will not have access to other students' grades without expressed consent. Similarly, any information about yourself that you share with your course instructor will not be given to anyone else without your permission. See <http://www.ucalgary.ca/policies/files/policies/privacy-policy-2011.pdf> for more information.

### **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 15 days** of first being notified of the grade.

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

Student Success Centre	<a href="http://www.ucalgary.ca/ssc/">http://www.ucalgary.ca/ssc/</a>
Student Wellness Centre	<a href="http://www.ucalgary.ca/wellnesscentre/">http://www.ucalgary.ca/wellnesscentre/</a>
Distress Centre	<a href="http://www.distresscentre.com/">http://www.distresscentre.com/</a>
Library Resources	<a href="http://library.ucalgary.ca">http://library.ucalgary.ca</a>

### **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 (Room 370 MacEwan Student 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/](http://www.ucalgary.ca/ombuds/) or email [ombuds@ucalgary.ca](mailto:ombuds@ucalgary.ca)

### **Student Union (SU) Information**

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

**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. The exam dates are firm and will not be altered.

January 10	Introduction (15 min) Activity: Foundational knowledge assessment (Basic biology: chromosomes, behavior of chromosomes, mitosis, meiosis, inheritance patterns, cell cycle regulation, transcription, translation, replication, gene) (Jillian Parboosingh) Gene structure, function and regulation (Bob Argiropoulos)
January 17	The human genome and its complexity: organization, variation and polymorphisms (Ryan Lamont). Activity: Variant databases, UCSC (Bob Argiropoulos)
January 24	Epigenetics, imprinting, X-inactivation, non-Mendelian genetics (Bob Argiropoulos) Activity: UCSC, Encode <b>Assignment #1 – due January 31</b>
January 31	Molecular genetic pathology: the consequence of small mutations (single nucleotide substitutions, small deletions, duplications, insertions) and their detection (Jillian Parboosingh /Ryan) Activity: Describing mutations (nomenclature), examples from molecular diagnostics <b>Assignment #2 GWAS literature critique – to be discussed February 7</b>
February 7	Roles of genes in complex disorders (Judy Chernos) Activity: GWAS studies - Literature critiques
February 14	Molecular genetic pathology: the consequence of dynamic mutations and their detection (Ryan Lamont/Jillian Parboosingh) Activity: examples from molecular diagnostics
February 21	<b>Reading week – no class</b>
February 28	Molecular genetic pathology: the consequence of copy number variation and pseudogenes and their detection (Ryan Lamont)  Activity: examples from molecular diagnostics <b>Assignment #3 – Due March 7</b>
March 7	Whole exome/genome/transcriptome sequencing (Ryan Lamont/Jillian Parboosingh) Activity: Risk assessment problems <b>Assignment #4 – Due March 14</b>

March 14	Mitochondrial genetics (guest: Tim Shutt) Activity: the challenge of diagnosing mitochondrial diseases (Ryan Lamont)
March 21	Determining pathogenicity: Use of Model organisms, patient cohorts and <i>in silico</i> analyses Activity: Computer database exercise.
March 28	Final in-class written assessment
April 4	Student presentations
April 11	Ethics (Judy)