

## CUMMING SCHOOL OF MEDICINE GRADUATE COURSE OUTLINE

| COURSE TITLE: Advanced Molecular Genetics |                                                                                                        |              |                                                                                                                                            |
|-------------------------------------------|--------------------------------------------------------------------------------------------------------|--------------|--------------------------------------------------------------------------------------------------------------------------------------------|
| Course                                    | MDSC 641.03                                                                                            |              |                                                                                                                                            |
| Pre/Co-Requisites                         |                                                                                                        |              |                                                                                                                                            |
| Faculty                                   | Cumming School of Medicine, Graduate Science Education                                                 |              |                                                                                                                                            |
| Instructor Name(s)                        | Jillian Parboosingh<br>Ryan Lamont<br>Bob Argiropoulos                                                 | Email        | <u>Jillian.parboosingh@albertapubliclabs.ca</u><br><u>Ryan.lamont@albertapubliclabs.ca</u><br><u>Bob.argiropoulos@albertapubliclabs.ca</u> |
| Office Location                           | ACH – Genetics &<br>Genomics                                                                           | Office Hours | By appointment                                                                                                                             |
| Instructor Email Policy                   | Please include MDSC 641 in the subject header. Instructor(s) will endeavor to respond within 48 hours. |              |                                                                                                                                            |
| Telephone No.                             | Dr. Parboosingh 403-955-2267                                                                           |              |                                                                                                                                            |
| TA Name, if applicable                    | NA                                                                                                     | Email        |                                                                                                                                            |
| Class Term, Days                          | Winter 2020; Wednesdays                                                                                |              |                                                                                                                                            |
| Class Times                               | 2:00 – 4:50 pm                                                                                         |              |                                                                                                                                            |
| Class Location                            | Alberta Children's Hospital, Lower Level B0-170 (Genetics Labs meeting room)                           |              |                                                                                                                                            |

# **COURSE INFORMATION/DESCRIPTION OF THE COURSE**

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.

## LEARNING RESOURCES/REQUIRED READING

Resources:

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)

# COURSE OBJECTIVES/LEARNING OUTCOMES

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.

## **CUT POINTS FOR GRADES**

This course adheres to the grading system outlined in the University of Calgary, Faculty of Graduate Studies Calendar. Grades of A+ and A are not distinguished in the calculation of GPAs. Percentage/letter grade conversion used for this course is as follows

| Grade | Grade Point<br>Value | Percentage<br>Conversion | Graduate Description                                                                                                                                     |
|-------|----------------------|--------------------------|----------------------------------------------------------------------------------------------------------------------------------------------------------|
| A+    | 4.00                 | 95-100                   | Outstanding                                                                                                                                              |
| А     | 4.00                 | 90-94                    | Excellent – superior performance showing comprehensive understanding of the subject matter                                                               |
| A-    | 3.70                 | 85-89                    | Very Good Performance                                                                                                                                    |
| B+    | 3.30                 | 77-84                    | Good Performance                                                                                                                                         |
| В     | 3.00                 | 72-76                    | Satisfactory Performance                                                                                                                                 |
| B-    | 2.70                 | 68-71                    | Minimum Pass for Students in the Faculty of Graduate Studies                                                                                             |
| C+    | 2.30                 | 63-67                    | All grades below 'B-" are indicative of failure at the graduate level<br>and cannot be counted toward Faculty of Graduate Studies course<br>requirements |

| Assessment Components: The University policy on grading related matters is outlined in the 2019-2020 |                                                    |             |                          |  |
|------------------------------------------------------------------------------------------------------|----------------------------------------------------|-------------|--------------------------|--|
| <u>Calendar</u> .                                                                                    |                                                    |             |                          |  |
| Assessment Methods                                                                                   | Description                                        | Weight<br>% | Due Date <u>and</u> Time |  |
| Assignment #1                                                                                        | The Human Genome and its Complexity<br>problem set | 10          | Jan. 29, 2020, 2 pm      |  |
| Assignment #2                                                                                        | Molecular Genetic Pathology problem set            | 10          | Feb. 12, 2020, 2 pm      |  |
| Assignment #3                                                                                        | Epigenetics problem set                            | 10          | Mar. 4, 2020, 2 pm       |  |
| Assignment #4                                                                                        | Structured Paper review                            | 10          | Mar. 25, 2020, 2 pm      |  |
| In-class participation                                                                               | In-class participation                             | 10          |                          |  |
| In-class problems                                                                                    | In-class written evaluation (all course material)  | 25          | Apr. 1, 2020, 2-5 pm     |  |
| Presentation                                                                                         | Presentation on approved topic                     | 25          | Apr. 8/15, 2020, 2-5 pm  |  |



#### ASSESSMENT AND EVALUATION INFORMATION

### ATTENDANCE AND PARTICIPATION EXPECTATIONS:

Students are expected to attend all classes and fully participate in discussions, being respectful of all points of view.

## **GUIDELINES FOR SUBMITTING ASSIGNMENTS:**

Assignments #1 - 4 may be hand written or typed. If hand written, writing must be legible and written in ink. Your name and student ID# must appear on the front page and all pages must be stapled together. Problem sets are to be handed in at the beginning of class on the due dates listed above.

### FINAL EXAMINATIONS: None

### **EXPECTATIONS FOR WRITING:**

Students are expected to write effectively to communicate their ideas and responses to questions. Excessive grammatical and typographical errors are minimally distracting and can mask a student's ability to demonstrate knowledge and understanding.

### LATE AND/OR MISSING ASSIGNMENTS:

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.

#### Is a passing grade on a particular component essential to pass the course as a whole? No.

|                            | COURSE TIMETABLE                                                 |                                   |                           |  |
|----------------------------|------------------------------------------------------------------|-----------------------------------|---------------------------|--|
| Course<br>Schedule<br>Date | Topic & Reading                                                  | Instructor                        | Assignments*<br>Due Dates |  |
| Jan. 15, 2020              | Human genome and its complexity<br>Foundational genetic concepts | Drs. Argiropoulos,<br>Parboosingh |                           |  |
| Jan. 22, 2020              | Molecular genetic pathology I: SNVs, inde                        | ls Drs. Lamont, Parboosingh       |                           |  |
| Jan. 29, 2020              | Molecular genetic pathology II: dynamic mutations                | Drs. Lamont, Parboosingh          | Assignment #1             |  |
| Feb. 5, 2020               | Molecular genetic pathology III: CNVs, pseudogenes               | Drs. Lamont, Parboosingh          |                           |  |
| Feb. 12, 2020              | Epigenetics, imprinting, X-inactivation                          | Dr. Argiropoulos                  | Assignment #2             |  |
| Feb. 26, 2020              | Clinical NGS-based tests<br>WES analysis                         | Drs. Lamont, Parboosingh          |                           |  |



| Mar. 4, 2020  | Mitochondrial Genetics                                               | Dr. Shutt (guest lecturer)      | Assignment #3 |
|---------------|----------------------------------------------------------------------|---------------------------------|---------------|
| Mar. 11, 2020 | Cancer Genetics                                                      | Dr. Box (guest lecturer)        |               |
| Mar. 18, 2020 | Cell free DNA: non-invasive prenatal testing, circulating tumour DNA | Dr. Parboosingh                 |               |
| Mar. 25, 2020 | Pharmacogenomics                                                     | Dr. Bousman (guest<br>lecturer) | Assignment #4 |
| Apr. 1, 2020  | Final in-class written assessment                                    | Dr. Parboosingh                 |               |
| Apr. 8, 2020  | Student presentations                                                | Drs. Argiropoulos, Lamont       |               |
| Apr. 15, 2020 | Student presentations                                                | Drs. Argiropoulos, Lamont       |               |

\*assignments are due at the beginning of class.

### INTERNET AND ELECTRONIC COMMUNICATION DEVICE INFORMATION

Cell phones must be turned off in class unless otherwise arranged with the instructor.

The use of laptop and mobile devices is acceptable when used in a manner appropriate to the course and classroom activities. Students are to refrain from accessing websites that may be distracting for fellow learners (e.g. personal emails, Facebook, YouTube). 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-communicationspolicy.pdf.

## MEDIA AND RECORDING IN LEARNING ENVIRONMENTS

### Media recording for lesson capture

The instructor may use media recordings to capture the delivery of a lecture. These recordings are intended to be used for lecture capture only and will not be used for any other purpose. Although the recording device will be fixed on the Instructor, in the event that incidental student participation is recorded, the instructor will ensure that any identifiable content (video or audio) is masked, or will seek consent to include the identifiable student content to making the content available on University approved platforms.

## Media recording for assessment of student learning

The instructor may use media recordings as part of the assessment of students. This may include but is not limited to classroom discussions, presentations, clinical practice, or skills testing that occur during the course. These recordings will be used for student assessment purposes only and will not be shared or used for any other purpose.

Media recording for self-assessment of teaching practices



The instructor may use media recordings as a tool for self-assessment of their teaching practices. Although the recording device will be fixed on the instructor, it is possible that student participation in the course may be inadvertently captured. These recordings will be used for instructor self-assessment only and will not be used for any other purpose.

# **Student Recording of Lectures**

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

# UNIVERSITY OF CALGARY POLICIES AND SUPPORTS

# ACADEMIC ACCOMMODATIONS

Students seeking an accommodation based on disability or medical concerns should contact Student Accessibility Services; SAS will process the request and issue letters of accommodation 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 their Instructor. The full policy on Student Accommodations is available at <a href="http://www.ucalgary.ca/policies/files/policies/student-accommodation-policy.pdf">http://www.ucalgary.ca/policies/files/policies/student-accommodation</a>

## **IMPORTANT INFORMATION**

Any research in which students are invited to participate will be explained in class and approved by the appropriate University Research Ethics Board

# 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). Further information for students is available on the Copyright Office web page (https://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 INTEGRITY

The Cumming School of Medicine expects intellectual honesty from its students. Course participants should be aware of University policies relating to Principles of Conduct, Plagiarism and Academic Integrity. These are found in the printed Faculty of Graduate Studies Calendar, or online under Academic Regulations in the Faculty of Graduate Studies Calendar, or online Studies Academic Regulations in the Faculty of Graduate Studies at Faculty of Graduate Studies Academic Regulations



## ACADEMIC MISCONDUCT

For information on academic misconduct and its consequences, please see the University of Calgary Calendar at <a href="http://www.ucalgary.ca/pubs/calendar/current/k.html">http://www.ucalgary.ca/pubs/calendar/current/k.html</a>

# EMERGENCY EVACUATION AND ASSEMBLY POINTS

Assembly points for emergencies have been identified across campus. The primary assembly points for South Campus (Health Science Centre (HSC); Health & Research Innovation Centre (HRIC); Heritage Medical Research Building (HMRB) and Teaching, Research and Wellness (TRW)) are:

- HSC and HMRB: HRIC Atrium (alternate assembly point is Parking Lot 6)
- HRIC: HMRB Atrium (alternate assembly point is Parking Lot 6)
- TRW: McCaig Tower (alternate assembly point is HMRB Atrium)

# 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 N of the Faculty of Graduate Studies Calendar. Students must follow the official process and should contact the Student Ombuds Office (<u>http://www.ucalgary.ca/provost/students/ombuds</u>) for assistance with this and with any other academic concerns, including academic and non-academic misconduct

# THE FREEDOM OF INFORMATION AND PROTECTION OF PRIVACY (FOIP) ACT

This course is conducted in accordance with the Freedom of Information and Protection of Privacy Act (FOIP) and 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. Assignments given by you to 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

## 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), <u>https://www.ucalgary.ca/wellnesscentre/services/mental-health-services</u> and the Campus Mental Health Strategy website <u>https://www.ucalgary.ca/mentalhealth/</u>"

# SUPPORTS FOR STUDENT LEARNING, SUCCESS, AND SAFETY

**Student Ombudsman**: The Student Ombuds' Office supports and provides a safe, neutral space for students. For more information, please visit www.ucalgary.ca/ombuds/ or email <u>ombuds@ucalgary.ca</u>



**Student Union:** The SU Vice-President Academic can be reached at (403) 220-3911 or suvpaca@ucalgary.ca; Information about the SU, including elected Faculty Representatives can be found here: <a href="https://www.su.ucalgary.ca">https://www.su.ucalgary.ca</a>

**Graduate Student's Association**: The GSA Vice-President Academic can be reached at (403) 220- 5997 or gsa.vpa@ucalgary.ca; Information about the GSA can be found here: https://gsa.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 <u>http://www.ucalgary.ca/security/safewalk</u>. 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.