

ACKNOWLEDGEMENT

UBC's Point Grey Campus is located on the traditional, ancestral, and unceded territory of the xwməθkwəyəm (Musqueam) people. The land it is situated on has always been a place of learning for the Musqueam people, who for millennia have passed on in their culture, history, and traditions from one generation to the next on this site.

COURSE INFORMATION

Course Title	Course Code Number	Credit Value
Human Genomics and Medical Genetics	MEDG420	3 credits

THIS COURSE WILL BE TAUGHT PREDOMINANTLY "IN PERSON".

PREREQUISITES

This is an upper level Medical Genetics course that requires a strong background in basic genetics - at least a 'B' grade in the prerequisites [BIOL 335](#) and/or [BIOL 338](#) - and a desire to engage in learning from independent reading, topic-specific presentations and class participation.

COREQUISITES

None.

CONTACTS

Course Coordinator	Contact Details	Office Location	Office Hours
Elizabeth M. Simpson, MSc, PhD	simpson@cmmt.ubc.ca	CMMT at BC Children's Hospital Research Institute	*
Course Instructor(s)	Contact Details	Office Location	Office Hours
Jan Friedman, MD, PhD, FAAP, FABMG, FCCMG, FRCPC	jan.friedman@ubc.ca	BC Children's Hospital Research Institute	§
Mahmoud Pouladi, MSc, PhD	mahmoud.pouladi@ubc.ca	BC Children's Hospital Research Institute	§
Colin J. D. Ross, MSc, PhD	colin.ross@ubc.ca	BC Children's Hospital Research Institute	§
Laura Arbour, MSc, MD, MSc, FRCPC, FCCMG	Laura.Arbour@viha.ca	Island Medical Program in Victoria	§

*Coordinator will be available by email. §Instructors will be available for ½ hour after their classes.

Otherwise, please contact instructors by email, using the course number (MEDG420) in the subject line. During their block, instructors will attempt to reply to email queries within 48 hours, but response time may be considerably slower when they are not active in the course instruction.

OTHER INSTRUCTIONAL STAFF

Course Teaching Assistant	Contact Details	Office Location	Office Hours
Kheireddin (Khair) Mufti	kmufti@popi.ubc.ca	BC Children's Hospital Research Institute	†

†Teaching Assistant will be available by email.

COVID SAFETY

You are required to wear a non-medical mask during our class meetings, for your own protection and for the safety and comfort of everyone else in the class. For our in-person meetings in this class, it is important that all of us feel as comfortable as possible engaging in class activities while sharing an indoor space. Non-medical masks that cover our noses and mouths are a primary tool for combating the spread of Covid-19. Further, according to the provincial mandate, masks are required in all indoor public spaces including lobbies, hallways, stairwells, elevators, classrooms and labs. There may be students who have medical accommodations for not wearing a mask. Please maintain a respectful environment <https://hr.ubc.ca/working-ubc/respectful-environment>.

COURSE STRUCTURE

The course consists of 4 blocks. Each block will focus on topics of current interest and active research within the broad field of Human Genomics and Medical Genetics, and each will be led by a different course instructor with specific expertise in the chosen topics. The purpose of this course is to serve as an entrée into these areas of scientific interest by providing essential background and engaging the class in an active 2-way discussion of relevant issues. Therefore, there is no textbook. Instead instructors will assign readings or other materials and expect considerable class participation in discussions that will be part of individual student evaluations. This class structure is incompatible with recordings and none will be made nor available.

Each class will be 1.5 hours (9-10:30), and include variable lecture and class discussion components in the different blocks. The instructor and guest speaker for Block 4 will be remotely located.

The specific topics to be covered in each block are listed below.

SCHEDULE OF TOPICS**Block 1: Identifying Genetic Variants That Cause Mendelian Disease (Jan Friedman)**

Wed Sep 8 – Genetic variants and human variation

Mon Sep 13 – Strategies for disease gene identification I: Approaches

Wed Sep 15 – Strategies for disease gene identification II: Technologies

Mon Sep 20 – Strategies for disease gene identification III: Application

Wed Sep 22 – Other applications of human genetic variation

Mon Sep 27 – Block 1 Guest, Shelin Adam, BSc, MSc, Genetic Counsellor, BC Children's Hospital

Block 2: Functional Genomics (Mahmoud Pouladi)

Wed Sep 29 – Introduction and overview of model systems

Mon Oct 4 – *In vivo* systems – Invertebrates: worms & flies

Wed Oct 6 – *In vivo* systems – Vertebrates: zebrafish, murine, and other models

Mon Oct 11 – No Class - Thanksgiving Day

Wed Oct 13 – *In vitro* systems – Immortalized, haploid, and hPSC models

Mon Oct 18 – Forward genetics/genetic screens

Wed Oct 20 – Block 2 Guest, Josef Penninger, MSc., PhD, Director, Life Sciences Institute, UBC

Block 3: Gene Therapy (Colin Ross)

Mon Oct 25 – What is gene therapy?

Wed Oct 27 – Vectors for gene therapy

Mon Nov 1 – Gene augmentation therapy

Wed Nov 3 – Genome editing therapy

Mon Nov 8 – Gene therapy project and discussions

Wed Nov 10 – No Class - Mid-Term Break

Mon Nov 15 – Block 3 Guest, Blair R. Leavitt MD, CM, FRCPC (Neurology), CMMT at BC Children's Hospital Research Institute

Block 4: Community Genomics/Genetics (Laura Arbour)

Wed Nov 17 – The application of Genomics/Genetics in the community-an overview

Mon Nov 22 – Genetics and Unique populations in Canada

Wed Nov 24 – Genetics/genomics and Indigenous communities

Mon Nov 29 – Newborn Screening-past, present and future

Wed Dec 1 – Direct to consumer genetic testing-benefit, entertainment, or peril?

Mon Dec 6 – Block 4 Guest, Keolu Fox, PhD, Anthropology, UC San Diego, La Jolla, CA, USA

LEARNING OUTCOMES

The objectives of this course are to introduce students to key concepts and methodologies fundamental to human genomics and medical genetics; to confer a basic understanding of current topics of interest, and for students to learn how to critically and effectively read scientific publications and understand principles of scientific investigation.

LEARNING ACTIVITIES

Student participation is an integral part of learning in this class, and participation will be evaluated in part by assessment of class discussions. Instructors may differ as to whether there are additional assignments to be completed for in-class discussion.

LEARNING MATERIALS

There is no textbook. Reading and other materials will be provided by the TA for each individual class on the UBC interactive learning interface (currently Canvas).

Students are expected to be able to work with spreadsheets. Software is available:

<https://it.ubc.ca/services/desktop-print-services/software-licensing/office-365-students>

ASSESSMENTS OF LEARNING

Breakdown of marks (per block)

Take Home Exams: 20 + 20 + 20 + 20 = 80% of total

Class Participation: 5 + 5 + 5 + 5 = 20% of total

There will four graded Take Home Exams – 1 per block, each worth 20% of the final course grade.

Students will be given a question, or questions, related to the topic of the block. Questions will be given out and are due exactly according to the schedule below. These exams must be undertaken individually, your answer should be your own original text. Do not copy any text from references in the body of your answers.

Format. There is no need to repeat the question(s) in your document. Your answer should be presented in single spaced Times New Roman or Calibri size 12 font, contained on not more than 4 pages with 2 cm borders on all sides. Any figures or tables must be included within the 4-page limit. References are to be cited numerically in the 4 pages of text, with full details of the references on separate page(s). Include your student number – but not your name – in the header at the top of each page of your exam.

Late submissions will lose 20% of the grade per day in the absence of a Clinician's note. Any requests for re-grading of exams will result in regrading of the entire assignment.

Any changes to the assessment plan will be discussed in class and a new syllabus would be available on CANVAS.

Exam Schedule

Block 1: Take Home Exam

Wed Sep 8 – Exam given out

Sun Sep 26 – Exam due before 9 pm

Block 2: Take Home Exam

Wed Sep 29 – Exam given out

Tue Oct 19 – Exam due before 9 pm

Block 3: Take Home Exam

Wed Oct 25 – Exam given out

Sun Nov 14 – Exam due before 9 pm

Block 4: Take Home Exam

Wed Nov 17 – Exam given out

Sun Dec 5 – Exam due before 9 pm

Participation in class will account for 20% of the final course grade.

Participation is evaluated not only on quantity (i.e. the frequency and/or duration of your participation) but also on its quality. For example, is the query, viewpoint of general interest, is the contribution informed, well reasoned and clearly presented/articulated, does the exchange elicit and advance discussion by others in the class? Participation will be evaluated in each class and an assessment amalgamated at the end of the block.

If you are unable to attend a class you will be allowed one absence if you inform the instructor before the start of the class. In this case, no grade will be entered and your participation will be averaged over the other classes. Additional days of illness will require a Clinician's note, otherwise a zero will be entered for participation that day. If you miss a class you will be responsible for contacting classmates to learn the material covered.

UNIVERSITY POLICIES

UBC provides resources to support student learning and to maintain healthy lifestyles but recognizes that sometimes crises arise and so there are additional resources to access including those for survivors of sexual violence. UBC values respect for the person and ideas of all members of the academic community. Harassment and discrimination are not tolerated nor is suppression of academic freedom. UBC provides appropriate accommodation for students with disabilities and for religious observances. UBC values academic honesty and students are expected to acknowledge the ideas generated by others and to uphold the highest academic standards in all of their actions.

Details of the policies and how to access support are available on [the UBC Senate website](#).

OTHER COURSE POLICIES

LEARNING RESOURCES

CANVAS will contain assigned materials, slides prior to the class, Take-Home Exams, as well as any supplemental material. Email the TA if you are not able to find these materials.

COPYRIGHT

All materials of this course (course handouts, lecture slides, assessments, course readings, etc.) are the intellectual property of the Course Instructor or licensed to be used in this course by the copyright owner. Redistribution of these materials by any means without permission of the copyright holder(s) constitutes a breach of copyright and may lead to academic discipline.

Students are not permitted to record classes.