

## ACKNOWLEDGEMENT

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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

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

## LOCATION: 1.410 LIFE SCIENCES CENTRE

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## PREREQUISITES

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One of [BIOL 335](#), [BIOL 338](#), or equivalent. A standing of 'B' or higher is strongly recommended in the prerequisite courses.

## COREQUISITES

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None.

## CONTACTS

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Course Instructor(s)	Contact Details	Office Location	Office Hours
Carolyn Brown	<a href="mailto:carolyn.brown@ubc.ca">carolyn.brown@ubc.ca</a>	LSI 5.506	*
Jan Friedman	<a href="mailto:jan.friedman@ubc.ca">jan.friedman@ubc.ca</a>	BC Children's Hospital Research Institute	
Elizabeth M. Simpson	<a href="mailto:simpson@cmmt.ubc.ca">simpson@cmmt.ubc.ca</a>		
Elizabeth Conibear	<a href="mailto:conibear@cmmt.ubc.ca">conibear@cmmt.ubc.ca</a>		

\*Instructors will be available for ½ hour after their classes at the UBC site. Please contact instructors by email at the email addresses listed, using the course number (MEDG420) as subject line. During their block, instructors will attempt to reply within 48 hours, but response time may be considerably slower when they are not active in the course instruction.

## OTHER INSTRUCTIONAL STAFF

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Teaching Assistant: Tiffany Carlaw [tiffany.carlaw@alumni.ubc.ca](mailto:tiffany.carlaw@alumni.ubc.ca)

## COURSE STRUCTURE

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The course consists of 4 blocks; each focused on a broad area of Human Genomics and Medical Genetics, and each with a different course instructor. There is no textbook, instead instructors will assign readings. Participation in class discussions based on these readings is an integral part of the class, and will be evaluated each class.

Each class will be ~ 1.5 hours (9-10:30), and include some lecture and considerable class discussion. The

extent of lecture will vary between instructors. A PDF of the slides for these lectures will be available on CANVAS before the class.

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

## SCHEDULE OF TOPICS

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### **Course Introduction (Carolyn Brown)**

Wed. Sept. 4 – The Human Genome: Genes and ...Junk ?

### **Gene mapping and identification in Mendelian disease (Jan Friedman)**

Mon Sept 9 – Genetic variants and human variation

Wed Sept 11 – Strategies for disease gene identification I: Approaches

Mon Sept 16 – Strategies for disease gene identification II: Technologies

[Wed Sept 18 – Guest Lecturer: Shelin Adam, Genetic Counsellor: Clinical genome-wide sequencing](#)

Mon Sept 23 – Strategies for disease gene identification III: Application

Wed Sept. 25 – Other applications of human genetic variation

### **Epigenetics and Epigenomics (Carolyn Brown)**

Mon Sept 30 – The Epigenome - Approaches

Wed Oct 2 – The Epigenome: DNA Methylation

Mon Oct 7– The Epigenome: Histone Modifications and Cancer

Wed Oct 9 – X-Chromosome Inactivation

[Mon Oct. 14 – Thanksgiving](#)

[Wed Oct 16 – Midterm Exam](#)

### **Gene Therapy (Elizabeth M. Simpson)**

Mon Oct 21 – What is Gene Therapy?

Wed Oct 23 – Augmentation Gene Therapy

Mon Oct 28 – Optogenetic Gene Therapy

Wed Oct 30 – Genome-Editing Therapy

Mon Nov 4 – Molecular Regulation of Gene Therapy

[Wed Nov 6 – GSC Tour \(570 West 7th Avenue - buzzer # 100\)](#)

[Mon Nov 11 – Remembrance Day](#)

### **Mechanisms of genetic disease; Personalized medicine (Liz Conibear)**

Wed Nov 13 – Genotype/phenotype correlations in cystic fibrosis

Mon Nov 18 – Using model organisms to find modifiers and prioritize candidates

Wed Nov 20 – Pharmacogenomics

Mon Nov 25 – Personalized medicine

Wed Nov 27 – Ethical considerations in personalized medicine

Date TBD (Dec. 3–18, 2019) – Final Exam on latter two blocks.

## LEARNING OUTCOMES

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The objectives of this course are to learn key concepts in human genomics and medical genomics; to critically and effectively read literature; and to learn how to independently research scientific topics in the literature.

## LEARNING ACTIVITIES

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Student participation is an integral part of learning in this class, and participation will be evaluated. There will be an excursion to the Genome Sciences Center (pending arrangements). Instructors may differ as to whether there are additional assignments to be completed for in-class discussion.

## LEARNING MATERIALS

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THERE IS NO TEXTBOOK.

Reading material as well as assignments for students will be assigned 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

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<b>Midterm:</b>	<b>30%</b>
<b>Final:</b>	<b>30%</b>
<b>Class Participation:</b>	<b>20%</b>
<b>Take Home Questions:</b>	<b>20%</b>

**There will be two exams, each worth 30% of the final course grade.**

The in-class midterm will evaluate material from Block 1 and Block 2. The final exam will evaluate material from Block 3 and Block 4.

In the absence of a textbook, you are responsible for the material in the articles assigned to the whole class. The key concepts from these papers will be discussed in class.

If you miss an exam you must provide a doctor's note and the examination will be rescheduled.

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

Participation is evaluated not only on quantity (ie whether you participate) but also on quality. For example, is the contribution well-thought out and clearly presented, does the contribution advance the class discussion? Participation will be evaluated in each class.

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 doctor'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.

**There will four graded Take Home Questions (THQs) – 1 per block, each worth 5% of the final course grade.**

Students will be given a set of questions related to an upcoming lecture. Brief written answers to this take-home question set are to be handed in before the start of the class (9 AM no exceptions) designated by the instructor. These are to be undertaken individually, and graded THQ's will be returned in class.

**Format.** There is no need to repeat the question in your document. Use a font size of 12, and at least 2 cm borders, and not less than single spaced. Your answer should be one page of text. Include references (with in-text citations), but on a separate page(s). Include your student number – but not your name – in the header.

Late submissions will lose 20% of the grade per day in the absence of a doctor's note.

Requests for re-grading of either exams or THQs 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.

## UNIVERSITY POLICIES

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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

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### LEARNING RESOURCES

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

### COPYRIGHT

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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.

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