

## 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, who for millennia have passed on their culture, history, and traditions from one generation to the next on this site.

## COURSE INFORMATION

Course Title	Course Code Number	Credit Value
Advanced Human Molecular Genetics	MEDG520	3

## PREREQUISITES

UBC BIOL335 (76% minimum) or equivalent genetics classes (evaluated by course coordinator).

Non-MEDG students interested in taking MEDG 520 must obtain permission from the coordinator. Email the coordinator a list of all upper level genetics courses that you have taken, your full transcripts & grades, and your CV; include your UBC student number.

## COREQUISITES

None.

## CONTACTS

Course Instructor(s)	Contact Details	Office Location	Office Hours
Dr. Carles Vilariño-Güell (course coordinator)	<a href="mailto:carlesvg@mail.ubc.ca">carlesvg@mail.ubc.ca</a> 604-827-1343	Djavad Mowafaghian Centre for Brain Health, 2215 Wesbrook Mall, Room 5639	All instructors will be available for approximately 30 minutes after class, or upon request by email
Dr. Stefan Taubert	<a href="mailto:taubert@cmmt.ubc.ca">taubert@cmmt.ubc.ca</a> 604-875-3860	BC Children's Hospital, CMMT, 950 W 28 <sup>th</sup> Ave, Room 2024	
Dr. Freda Miller	<a href="mailto:freda.miller@msl.ubc.ca">freda.miller@msl.ubc.ca</a> 604-827-0863	Michael Smith Laboratories, 2125 East Mall, Room 305	
Dr. Elizabeth Conibear	<a href="mailto:conibear@cmmt.ubc.ca">conibear@cmmt.ubc.ca</a> 604-875-3898	BC Children's Hospital, CMMT, 950 W 28 <sup>th</sup> Ave, Room 2108	
Dr. Inanc Birol	<a href="mailto:ibirol@bcgsc.ca">ibirol@bcgsc.ca</a> 604-707-5800	BC Cancer Agency, Genome Sciences Centre 570 W 7th Ave, #100	
Dr. Kelly McNagny	<a href="mailto:kelly@brc.ubc.ca">kelly@brc.ubc.ca</a> 604-822-7824	Centre for Blood Research 2222 Health Sciences Mall	
Dr. Peter Lansdorp	<a href="mailto:plansdor@bccrc.ca">plansdor@bccrc.ca</a> 604-675-8135	Terry Fox Laboratory, BC Cancer Agency, 675 West 10 <sup>th</sup> Avenue	
Dr. Cornelius Boerkoel	<a href="mailto:nboerkoel@bcchr.ca">nboerkoel@bcchr.ca</a> 604-875-2157	BC Children's Hospital, 950 W 28 <sup>th</sup> Ave	

## OTHER INSTRUCTIONAL STAFF

Teaching Assistant: Emilie Theberge, [Emilie.Theberge@bcchr.ca](mailto:Emilie.Theberge@bcchr.ca)

## OVERVIEW

MEDG 520 (3 credits) is a core course for graduate students in Medical Genetics and covers advances in human molecular genetics and genomics. It is recommended that students complete MEDG 520 prior to MEDG 530; however, in certain circumstances (such as a January-start students) the courses may be taken in reverse order. All enrolled students are expected to have a strong background in the principles and fundamentals of genetics and genomics.

Please note that the concepts taught/learned in MEDG 520 are a key component of the comprehensive exam that MEDG students take prior to transferring to PhD studies and advancing to PhD candidacy. Approximately 50% of the comprehensive exam will be an examination of these concepts (see: <https://med-fom-medgen.sites.olt.ubc.ca/files/2021/02/Guidelines-for-the-PhD-Comprehensive-Examination-Feb-2021-final.pdf>).

## COURSE STRUCTURE

The course consists of eight blocks covering topics in human molecular genetics and genomics with individual faculty instructors for each block (see schedule). Class format may vary between blocks. The course includes a mid-term mark based on presentations and a final written exam.

## SCHEDULE OF TOPICS

The class meets Tuesdays and Thursdays in two sections: Section 1 from 8:30 am to 10:00 am, and Section 2 from 10:30 am to 12:00 pm. During the intro class, students are assigned either to Section 1 or Section 2. They then remain in their assigned section for all of MEDG 520. Please note that some students are pre-assigned to a section due to scheduling constraints.

Block	Instructor	Topic	Date(s)	Location
Intro #1	Carles Vilariño-Güell	Intro class (10:30-11:30am for all students)	Sept 7	Zoom
Intro #2	Stefan Taubert	Grad School 101 (10:30-11:30am for all students)	Sept 9	Zoom
1	Stefan Taubert	Non-Mendelian inheritance	Sept 14, 16	SCRF 1020
2	Carles Vilariño-Güell	Genetic Variability and Gene Mapping	Sept 21, 23, 28	SCRF 1020
	Truth and Reconciliation		Sept 30	
3	Freda Miller	Stem cells and regeneration	Oct 5, 7	SCRF 1020
4	Elizabeth Conibear	Membrane transport processes	Oct 12, 14, 19	SCRF 1020
5	Inanc Birol	Genomics & Bioinformatics	Oct 21, 26, 28	SCRF 1020

6	Neal Boerkoel	Clinical Genomics	Nov 2, 4, 9	SCRF 1020
Midterm break			Nov 11	
7	Peter Lansdorp	Genome Instability, Aging & Senescence	Nov 16, 18, 23	SCRF 1020
8	Kelly McNagny	Immunogenetics	Nov 25, 30, Dec 2	SCRF 1020

**Mid-term mark:** Based on participation and concept presentations, a composite mark reflecting work from the first four blocks will be provided the week of October 25<sup>th</sup>

**Final exam:** Thu Dec 9<sup>th</sup>, 10am-1pm. Open-book online written exam. Eight out of sixteen questions must be answered, one from each block, within a fixed space limit.

## LEARNING OUTCOMES

The objectives of this course are to:

1. Learn key concepts in human genetics and genomics.
2. Critically and effectively read original research papers.
3. Learn how to independently research and present scientific topics using published literature.

## LEARNING ACTIVITIES

This course is conducted in a student-led discussion format, in which the students learn to teach themselves with guidance from instructors. The course employs interactive student-led discussion of key research concepts and paper(s) assigned prior to each class, followed by an instructor-led presentation to emphasize key concepts. The first class in each block may include an introductory lecture covering the relevant topic.

## LEARNING MATERIALS

Reading material as well as assignments for students will be assigned by the TA for each class on UBC's interactive learning interface, Canvas (<https://canvas.ubc.ca>).

## ASSESSMENTS OF LEARNING

The course will be graded based on:

- Class participation (40%)
- Student presentations of assigned concepts (30%)
- A final exam (30%)

## 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 and 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](#).

Covid-19 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 teaching labs. There may be students who have medical accommodations for not wearing a mask. Please maintain a respectful environment. [UBC Respectful Environment Statement](#).”

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## LEARNING ANALYTICS

Learning analytics includes the collection and analysis of data about learners to improve teaching and learning. In this course, it is planned to:

- View overall course progress
- Acquire personalized feedback via a mandatory questionnaire at the conclusion of the course

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## LEARNING RESOURCES

Source material required to cover content in this course will be shared on Canvas and/or available through PubMed or the UBC library.

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## COPYRIGHT

All materials of this course (course handouts, lecture slides, assessments, course readings, etc.) are the intellectual property of the course instructors 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.

**Recording of this class by students is not permitted.**

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## LOCATION OF CLASSES

MEDG 520 is expected to take place primarily in-person in Room 1020, at the Nevile Scarfe building, 2125 Main Mall, V6T 1Z4 with the exception of the first two lectures (Sept 7 & 9) which will be delivered online over zoom. However, due to the current and increasing incidence of the COVID-19 Delta variant it may be necessary to return to online teaching for the entirety of the course. For further information on COVID-19 and return to on-campus activities, see:

<https://students.ubc.ca/campus-life/returning-to-campus>

<https://covid19.ubc.ca/>

Students with disabilities are encouraged to reach out to the [Centre for Accessibility](#). Please email the course coordinator if you need further information.