

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) | carlesvg@mail.ubc.ca 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 | taubert@cmmt.ubc.ca 604-875-3860 | BC Children's Hospital, CMMT, 950 W 28 th Ave, Room 2024 | |
| Dr. Freda Miller | freda.miller@msl.ubc.ca 604-827-0863 | Michael Smith Laboratories, 2125 East Mall, Room 305 | |
| Dr. Elizabeth Conibear | conibear@cmmt.ubc.ca 604-875-3898 | BC Children's Hospital, CMMT, 950 W 28 th Ave, Room 2108 | |
| Dr. Kelly McNagny | kelly@brc.ubc.ca 604-822-7824 | Centre for Blood Research 2222 Health Sciences Mall | |
| Dr. Peter Lansdorp | plansdor@bccrc.ca 604-675-8135 | Terry Fox Laboratory, BC Cancer Agency, 675 West 10 th Avenue | |
| Dr. Cornelius Boerkoel | nboerkoel@bcchr.ca 604-875-2157 | BC Children's Hospital, 950 W 28 th Ave | |
| Dr. Adi Steif | adi.steif@ubc.ca 604-707-5900 | Genome Sciences Centre, 570 W. 7th Ave, Office 1.103.1 | |

OTHER INSTRUCTIONAL STAFF

Teaching Assistant: Martin Wong, Mtw17@student.ubc.ca

OVERVIEW

MEDG 520 (3 credits) is a core course for Genetic Counselling and Medical Genetics graduate students, 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 MSc defense and 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/2022/04/Guidelines-for-Masters-Thesis-and-Defence-1.pdf> & <https://med-fom-medgen.sites.olt.ubc.ca/files/2022/02/Guidelines-for-the-PhD-Comprehensive-Examination-Feb-25-2022.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, however it should be structured around three ~10 min student presentation + ~10 min Q&A followed by a 30min lecture.

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) | Sep 6 | Zoom |
| Intro #2 | Stefan Taubert | Grad School 101 (10:30-11:30am for all students) | Sep 8 | Zoom |
| 1 | Stefan Taubert | Non-Mendelian inheritance | Sep 13, 15 | LSC 1410 |
| 2 | Carles Vilariño-Güell | Genetic Variability and Gene Mapping | Sep 20, 22, 27 | LSC 1410 |
| 3 | Freda Miller | Stem cells and regeneration | Oct 4, 6, 11 | LSC 1410 |
| 4 | Elizabeth Conibear | Genetic variants and protein function | Oct 13, 18, 20 | LSC 1410 |

| | | | | |
|---------------|----------------|---|-------------------|----------|
| 5 | Kelly McNagny | Immunogenetics | Oct 25, 27, Nov 1 | LSC 1410 |
| 6 | Neal Boerkoel | Clinical Genomics | Nov 3, 8 | LSC 1410 |
| Midterm break | | | Nov 10 | |
| 6 | Neal Boerkoel | Clinical Genomics | Nov 15 | LSC 1410 |
| 7 | Peter Lansdorp | Genome Instability, Aging & Senescence | Nov 17, 22, 24 | LSC 1410 |
| 8 | Adi Steif | Statistics, probability and data analysis | Nov 29, Dec 1, 6 | LSC 1410 |

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 24th

Final exam: Thu Dec 13th, 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 as per current UBC policy (<https://srs.ubc.ca/covid-19/covid-19-safety-plans/>), 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. There may be students who have medical accommodations for not wearing a mask. Please maintain a respectful environment. [UBC Respectful Environment Statement](#).”

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

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.

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.

LOCATION OF CLASSES

MEDG 520 is expected to take place primarily in-person in Room 1410, at the Life Sciences Centre (LSC), 2350 Health Sciences Mall, BC V6T 1Z3 with the exception of the first two lectures (Sept 6 & 8) which will be delivered online over zoom.

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