

MEDG 530

HUMAN GENETICS

W2025/2026

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Faculty

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Block 1 (Jan 6- Jan 29)

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Teaching Assistant

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All rooms are located in the Shaughnessy Wing of BC Women's Hospital

Organization of the Course:

MEDG 530 employs problem-based learning. The class will generally meet in small tutorial groups each Tuesday and Thursday morning from 08:30-10:30.

Classes for UBC start January 05, 2026 so the first MEDG 530 tutorial session is Tuesday, January 06, 2026 and the last tutorial will be Thursday April 2, 2026.

Oral examinations will take place on April 7 & 9, 2026.

In problem-based learning, **students work cooperatively, and the students themselves are primarily responsible for their own learning.** The tutorials are designed to provide a context that will encourage students to

- Employ effective problem-solving skills;
- Apply previously-learned knowledge;
- Synthesize new information with prior knowledge;
- Develop self-directed learning skills;
- Build an organizational framework for further learning;
- Provide motivation for further learning; and
- Evaluate what has been learned.

This is an advanced graduate course in human genetics. Students must have successfully completed at least two one-term undergraduate genetics courses and a half-year graduate course (MEDG 520) is generally required before enrolling in MEDG 530. **Knowledge of human genetics equivalent to that provided in a full-year senior undergraduate genetics course is necessary to understand the cases in MEDG 530. *Students who need to review basic genetic principles should read the relevant sections in a standard human genetics textbook before each new case.***

The course is divided into six blocks. Each of the six blocks includes 4 classes devoted to a case. Each case will be introduced during the last 15-20 minutes of the class preceding the start of a new block, so that students will know the topic being discussed when the block begins. This will give all students the opportunity to read generally about the topic so that they can participate fully in the discussion beginning with the first full session of the block. During each session, students will be given additional information about the case and will identify important problems that it presents.

As the students assess and discuss a case, they will create a list of learning issues needed to understand the problems fully. Each group will identify its most important learning issues and assign them to its members to learn about. Some overarching learning issues may be assigned to all class members; more limited learning issues may only be assigned to one or two students. Group members will then use the scientific literature and other available resources to learn about their assigned issues before the next class. When developing their learning issues, ***students should strive to understand the important genetic principles that underlie the problem being presented.*** In general, students are not expected to learn the clinical features or management of the diseases used in the cases, unless these are adopted as specific learning issues.

In the tutorial sessions on each case, students will review what they have learned with each other and discuss how it relates to the previously-identified problems. As additional information about the case is provided, students will interpret this in terms of what they have learned. At the end of each case, students will be given the objectives the case was designed to teach. This will permit students to make certain that they have learned what was expected and to “fill in the gaps,” if necessary.

Student presentations should be brief and focused on the learning issues. An individual student’s presentation should not take more than 5-10 minutes, although the subsequent discussion it evokes will often take longer. The most important considerations in determining each student’s participation mark will be the student’s overall contribution to the group’s discussion and learning, the quality of the information presented, and its relevance to the problem at hand. Class participation is essential. ***Every student is expected to participate proactively in the discussion in every class.*** Students who contribute minimally (e.g. providing only a cursory overview of their assigned topic such as might be gleaned from Wikipedia) will be awarded fewer marks than students who contribute actively and demonstrate evidence of detail-oriented thought.

The course website on UBC Canvas will be used to post the cases for discussion, as well as to post exams set by the Course Coordinator.

General Course Objectives: Students who complete this course will be able to

- Use the published scientific literature and online resources to find information on particular problems in human genetics;
- Read and understand pertinent scientific literature and interpret it in the context of a specific problem;
- Distinguish between information that is relevant to a problem and information that is not relevant;
- Determine what is firmly established and what is only partly known, conjectural, or unknown in a particular area; and
- Use the information obtained to formulate a reasonable approach to solving a scientific problem in Human Genetics, and to explain the rationale for the approach chosen.

Grading: There will be two examinations in the course. The midterm exam will comprise 25% of the total course mark. The final exam will comprise 50% of the total course mark. The other 25% of the total mark will be for class participation. Each student's participation will be scored for each class on a 5-point scale, as follows:

0	Absent
1	Present but did not participate meaningfully in discussion
2	Minimal participation (e.g. returned answers based only on textbooks or Wikipedia entries without showing any understanding of what they meant in the context of the case being discussed)
3	Participated but contributed little to discussion (e.g. comments were superficial, and/or focused primarily on an aspect of the problem that was tangential to the main point)
4	Participated fully and contributed well to the discussion
5	Outstanding participation (e.g. integrated concepts, distinguished material that was relevant to the problem from irrelevant material, clarified and provided perspective to material presented by others when appropriate)

Both the midterm and final are take-home examinations that will be posted on UBC Canvas about one month before the due date. In addition, the final exam will include a 10 minute in-person presentation on their paper to the course coordinator and teaching assistant. ***Students must work alone on the exams***, although they may use the scientific literature, textbooks, or any other published source of information to help formulate answers. The answers must be typed, double-spaced, in a 12-point font with 2.5 cm margins to fit standard 21.6 x 27.9 cm (8.5 x 11 inch) paper. The answers cannot exceed the stated page limits given for each question. References should be included as appropriate at the end of the answer for each question but are not counted in the page limit.

Midterm Exam DUE: Friday, 13 February 2025

Written Final Exam DUE: Thursday, 2 April 2025

Late Penalty:

Penalty for late submission of a midterm or final exam is 10% of the total mark per day late. The penalty accrues instantaneously, so an exam that arrives at 08:30 and 59 seconds is not late, whereas an exam that arrives at 08:31 and 0 seconds is late. The penalty accrues irrespective of E-mail server failures, delayed receipt, etc. Don't be late.

Extensions on Deadlines:

Extensions must be requested at least 48 hours in advance of the deadline via a single E-mail of no more than 250 words that lays out explicitly the extenuating circumstances that justify such a request. The student must specify how much extra time he or she is requesting, and must provide signed external documentation (e.g. doctor's note) to support claims of sudden illness. The course coordinator cannot meet personally with students to discuss extenuating circumstances. The course coordinator's ruling is final and no correspondence can be entered into.

There are no extensions on extensions. If an exam or paper is submitted **after** a previously agreed-upon extended deadline has expired, the penalty shall be 10% of the total mark per day late according to the **original (non-extended)** deadline. Thus, a student who is granted a 24-hour extension and submits his or her paper 23 hours, 59 minutes and 59 seconds after the original deadline would receive no time-related penalty, whereas a student who is granted a 24-hour extension and submits his or her paper 24 hours, 0 minutes and 1 second after the original deadline would receive a 20% penalty.

So don't be late, and if you have to be late for an agreed-upon reason, don't be later.

Teaching Assistant:

The Teaching Assistant is available to help students understand difficult issues raised by the cases, suggest resources to understand learning issues, and gain knowledge in areas in which the student needs additional work.

Grading by the TA and Course Coordinator of Your Midterm and Final:

We mark hard.

That is to say, students are expected to think and write the answers to their questions as though they were submitting a short review for publication, with the additional expectation that they will develop a line of reasoning and show their understanding of the topic within the content of their essay. Simply citing someone else's article that makes your argument for you is not enough. Many students who have not previously taken logic and/or philosophy courses find this type of writing to be exceptionally challenging. All students should review the material available at UBC's Science Writing Resources for Learning (ScWRL) website: <https://scwrl.ubc.ca/>

Your marks in MEDG 530 are to be treated as formative evaluation, not summative evaluation. If you choose to appeal a grade you have received on your written work, you must first discuss your written work one-on-one with your Research Supervisor, in detail. During this discussion the two of you must also discuss, in detail, the comments from the TA and Course coordinator. This process is to encourage you to think deeply about your writing, referencing and reasoning. If your supervisor agrees with you that you have been marked too hard, then your supervisor is welcome to contact the Course Coordinator to discuss an appeal.

Tutorial Groups:

The class is divided into three tutorial groups – group memberships will be posted in Canvas, and students will remain with their tutorial group for the duration of the class.