

Department of Biology Course Outline

SC/BIOL 4285 3.00 Human Molecular Genetics
Winter 2020

Course Description

This course covers the application of genetic and molecular biological techniques to study human diseases and other related areas and discusses ethical concerns that might arise from this research.

Prerequisites

Prerequisite or co-requisite: SC/BIOL 3130 3.00

Course Instructors and Contact Information

Course Instructor: Dr. Tanya Da Sylva

Office: 151A Farquharson

Office hours As posted on Moodle or by appointment

- Office hours are times when you can drop-in (no appointment necessary) and ask questions or discuss course matters.

Email: dasylva@yorku.ca

Emails MUST have BIOL 4285 in the subject line. Emails must also include your name and student number. Expect an average response time of 3 business days. Email will not be checked on weekends or holidays. Please see the *Course Policies* section for more email guidelines.

Email is not a substitute for office hours. Some questions and/or concerns are better addressed in person and you will be asked to come to office hours or make an appointment with me.

Schedule

Fridays, 11:30 am – 2:30 pm in SLH C

Evaluation

The final grade for the course will be based on the following items weighted as indicated+:

Participation (in-class and on Moodle; throughout term) _____	10%
Point-Counterpoint Part 1 (February 28 th) _____	10%
Point-Counterpoint Part 2 (March 13 th) _____	5%
Innovation of the Decade (April 3 rd) _____	20%
Midterm (in class; February 14 th) * _____	20%
Final Exam (TBD; announced by Registrar) _____	35%

**If you do better on your final exam than the midterm, 5% weight from the midterm will be transferred to your final exam.*

+Final course grades may be adjusted to conform to Program or Faculty grades distribution profiles

Important Dates

Classes Start: Monday, January 6th

Winter Reading Week (NO CLASS): February 15th – 21st

Drop Date: March 13th

Last Day of Classes: April 5th

Course Withdrawal Period: March 14th – April 5th (course appears on your transcript without a grade and with a “W” notation)

Exam Period: April 7th – 25th (inclusive)*

*** It is your responsibility as a student to ensure that you are available to sit for examinations during the entire exam period.**

NOTE: Important dates may change to verify and for additional important dates such as holidays, refer to the “Important Dates” section of the Registrar’s Website at <http://www.yorku.ca/yorkweb/cs.htm>

Resources

Textbook: We will not be using a textbook or course pack. You will be provided with a list of mandatory and suggested material through Moodle. All material will be freely accessible online or available through York Libraries. Material will be provided and updated as the course progresses. Please check Moodle often.

Course Moodle Site: Required and suggested course material including readings and videos will be posted to Moodle. Announcements, grades and other course information will also be there. Moodle will be updated frequently, please check it often.

iClicker (REEF): iClicker is a student response system offered for free to York University students. We will occasionally use iClicker to poll the class or to answer questions. Registration, and **further instructions are on Moodle**. In order to use iClicker, you will need a charged laptop, tablet or smartphone. As we are only using iClicker occasionally, if you do not have any iClicker capable device with you when we are using it there will be an alternate means of recording your participation.

Learning Outcomes

- The main objective of the course is to develop an understanding of the molecular genetic and genomic techniques we use to study human variation including genetic diseases. As well as understanding the impact the field of human genetics has on society.
- You will: learn how the study of the human genome has changed over the last few decades and discuss emerging new techniques; investigate how the genetic contribution to human traits including diseases is determined; and discuss the implication of genetics and genomics on basic science and on the future of medical treatment and disease prevention.
- You will discuss ethical concerns raised by research in human genetics and investigate the impact of genetic discoveries and techniques on individuals and communities.

Course Content

You will be provided with a list of required and suggested readings or other material for each section of the course on Moodle. **You are expected to review the material before we start covering the topic in class.** Some tips on how to read articles are provided on Moodle. Reviewing material before class is meant to prepare you to actively participate in class.

In class we will spend most of our time actively engaging with material. You will be engaging in activities that promote analysis, synthesis of the course content, application to 'real-life' experiences and reflection on your learning process. Sometimes this will take the form of individual work, but often you will be participating in small group discussions or analysis.

In class I will provide you with the information and tools that will help you navigate the topic. However, **most of class time will be dedicated to activities and discussion.** You are expected to attend class and participate in all activities.

The course is divided into five units (more detail provided on Moodle):

- Unit 1 – The Genetic Revolution
- Unit 2 – Methods in Genetics, Transcriptomics and Epigenomics
- Unit 3 – Techniques in Medical Genetics
- Unit 4 – Genetic Contributions to Human Health and Disease
- Unit 5 – Genetic Engineering

Experiential Education and E-Learning

Experiential education: You will gain hands-on skill development in teamwork and scientific communication. You will go through case studies and be expected to apply course concepts to real world issues. Throughout the term you will be asked to reflect on your learning and connect course concepts to your own experiences.

Other Information

Lecture Recordings. I may occasionally provide lecture recordings for you, not all lectures will be recorded.

If you wish to make your own recordings, audio recording (ONLY audio) of the lecture is permitted. Recordings can only be shared with students enrolled in the course and section the recording was made of (your classmates). NO PHOTOS or VIDEOS.

Lecture Notes. I may post outlines of the lecture to Moodle before or after the lecture. These will not be available for all lectures. A summary of key points and/or learning objectives will be posted to Moodle for each unit when we complete the unit.

Class time will mostly be dedicated to discussion and small group activities, not lecturing. Therefore, both lecture recordings and outlines will be of limited use.

Class Attendance. You need to attend all lectures, and actively participate in order to do well in the course. The discussions in class are critical for understanding the material. Participation in class and on Moodle is also worth 10% of your grade. Please see the *Course Policies* section below for the *Participation Policies*.

Course Material. This course will be taught using the scientific literature (primary and secondary sources) and various online resources. The list of required and optional material for each part of the course will be posted to Moodle. All material will be freely accessible online or through York Libraries.

Tests. The midterm and final may consist of multiple-choice, short, or long answer questions. Your comprehensive final will take place during the exam period and will have the same type of questions as the midterm and may have essay questions.

You will be responsible for the required material as listed on Moodle and the lecture material. Test questions may cover material not covered in class. Please see the *Course Policies* section below for information on missed tests.

Point-Counterpoint Assignment. This assignment is meant to build your critical thinking and argumentation skills while deepening your understanding of the impact the human genetics field has on society.

You will be given a question and will choose whether to argue for or against. The number of students for and against will be limited, and a form will be placed on Moodle allowing you to choose a side (first come, first served). You will research and analyze information about the topic and compose a written argument in support of your position. This assignment will be done in two parts.

In Part 1 (10%) of the assignment you will submit a written argument, following the guidelines, provided. Your submission will be made both to me (for marking) and posted in a forum on Moodle accessible to the class. In Part 2 (5%) you will revise your argument, updating your paper in response to some of the points raised by the opposing side (which you can view in the forum). This written revision will be submitted only to me (for marking). Please read the assignment outline for more details.

Innovation of the Decade Assignment. You will enhance your understanding of emerging techniques and issues in human genetics by writing a report on a major innovation in the field during the last decade (2010-2019). You will be allowed to choose your own topic.

Your report will illustrate why this innovation stands out amongst all others of the decade and will include a review of the techniques and research in the field that supported the development of the innovation. In drawing connections between past techniques/research and the innovation you will strengthen your understanding of the scientific process and knowledge building underlying human genetics research.

You will also discuss the questions, gaps in the field and/or other needs the innovation is meant to address. Your discussions will include the current and potential impact of the innovation on the field of human genetics/genomics and on individuals and society.

Overall this paper will strengthen your ability to draw connections between ideas, synthesise material and evaluate changes to a field of research while also deepening your understanding of course content.

Course Policies

Grading and Policies for Missed Tests and Assignments

Grading: The grading scheme for the course conforms to the 9-point grading system used in undergraduate programs at York (for a full description see the York University Undergraduate Calendar – <http://calendars.registrar.yorku.ca/2014-2015/academic/grades/index.htm>)

Missed Midterm/Exam – General Policies: Students must have an **urgent reason** for missing a course test, such as illness, compassionate grounds, etc., Supporting documentation (e.g., attending physician's statement) may be required.

- **It is YOUR responsibility to get to class on time.** Plan to be on campus early on test days; transit and/or traffic issues happen but you cannot be given extra time to write. Plan to be at exams early.
- **Bad weather will not postpone a test unless York officially closes campus.**
- Valid and appropriately detailed documentation supporting the events preventing your attendance may be required.
- **NOT all situations will be accommodated**, meaning that a zero will be assigned on the missed test.

Midterms: You **MUST** fill in the missed test form on Moodle **within five business days** of missing the test (the sooner the better). The form will explain any documentation necessary.

- **One make-up will be scheduled for each midterm.** If you have a valid reason for missing both the original midterm and the make up midterm then the weight of the missed test(s) will be transferred to your final exam. Make up tests may occur anytime before the end of the last class and may not be the same format as the original test. Further extensions or accommodation will require you to submit a petition to the Faculty.

Final Exam: *It is your responsibility as a student to ensure that you are available to sit for examinations during the entire exam period.*

- If you miss the final exam for a valid reason **you must request deferred standing.** See myacademicrecord.students.yorku.ca/deferred-standing for additional information.
- To request deferred standing, **you must complete a Deferred Standing Agreement Form and submit it along with supporting documentation** to me (T. Da Sylva) within 5 business days of the missed exam. Scanning the forms and emailing them to is preferred. **Requests submitted after this time will be denied and you must formally petition**
- If you were ill, you **MUST have your doctor fill out the Attending Physician's Statement** (make sure you are using the current version form; no other versions will be accepted). Please contact me about required documentation for non-medical circumstances.
- The date/time of the deferred exam is at my discretion. Deferred exams take priority over work for other courses. **If you miss the deferred exam you will have to formally petition for further accommodation.**
- If I deny the deferred standing request, you must submit a petition. Denied petitions will result in a zero on the final exam.
- Religious accommodations must be arranged at least 3 weeks prior to the exam (see general course policies below for guidelines).

Incomplete/Late Course Work Policies

In general, **extensions for any component of the *Point-Counterpoint* or *Innovation of the Decade* assignments will not be granted.** You are encouraged to plan ahead; do not leave completion to the last minute! Individuals with extenuating circumstances can contact me. All late submissions, without permitted extensions, will be penalized as noted in the assignment outline.

Participation Policies

Class attendance is mandatory; you are expected to attend class and participate actively, which requires you to come prepared. You are expected to engage respectfully and contribute meaningfully to all discussions and activities (in class or online).

Participation will be recorded multiple ways, including but not limited to, worksheets or reflections that are handed in, iClicker responses, and the taking of attendance. You cannot sign work or log attendance for someone else. That would be a violation of academic honesty and will be treated as such.

Missing class, arriving late or leaving early occasionally is often inevitable. There are twelve classes throughout the semester. One of the twelve classes will also be the day of the midterm (see missed midterm policies above). Therefore, there are eleven classes where participation will be recorded. You may miss up to three without penalty, this “miss” includes not being in class when participation was recorded for any reason (missed class, arrived late, left early). That means **you must attend and actively participate in at least 8 classes for full participation marks. No extra credit or make-up participation will be provided.**

I understand that there may be special circumstances that interfere with regular class attendance. Please discuss circumstances and accommodation needs with me (Dr. T. Da Sylva; dasylva@yorku.ca). Participation is worth 10% of your final grade.

Test Marks & Reviewing Tests

Test grades will be posted to Moodle when they are available. A test viewing time will be arranged for each test and announced via Moodle.

- The Midterm and Exam will not be handed back, but you will have opportunities to review your tests. These dates will be posted to Moodle, please do not email asking about viewing, a time will be announced. If you cannot attend the posted dates you may have to wait until after the final exam to view your test.

Re-marking of Tests or Assignments

If you believe a written answer on a test or assignment was marked incorrectly you must email me a **request for re-marking within 1 week** of the test/assignment being made available to you. You must put the test/assignment name and “re-marking request” in the subject line of the email.

Only answers in ink are eligible for remarking. Multiple choice questions cannot be re-marked.

Re-marking can result in the mark being raised, confirmed, or lowered.

- Your **re-marking request must include a written rationale** providing **academic grounds** for the re-marking. Your rationale should show why you believe your original marker was **factually wrong** in attributing this grade. You must detail how your answer was factually right and where the errors were in the marking.
 - Statements such as “this mark doesn’t reflect how hard I studied” or “I really know the material well and I should have a better mark” do not show academic merit.
 - You cannot compare your answers to other students’ answers. Your answer must have merit ON ITS OWN. For instance, statements like “my friends gave a similar answer and they got more marks” is not an acceptable reason for a re-marking.

- You will be told if your re-marking request is denied, as soon as possible. If your re-marking request is accepted, it may take some time for the re-marking to be completed. You may not hear about your re-marking until final grades are released.

To be fair and consistent to the entire class, **individual grades are not negotiable**. There are no 'extra credit' assignments, and grades are not "curved" or "bumped".

Email Policy & Conduct

You can reach me (T. Da Sylva) via email at dasyilva@yorku.ca. Expect an **average response time of 3 (working) days**, not including weekends or holidays. **Do not use the Moodle email (messaging) function** to contact me.

Email is not a substitute for attending drop-in (office) hours. You are also encouraged to ask questions in lectures and tutorials.

Due to the volume of email I receive and the concepts we cover in the course it may not be possible to address your question over email. Please read the following for **general guidelines and policies when emailing**:

- Emails from addresses other than your @my.yorku.ca address may be filtered as junk and not get a reply.
- **Please make your subject line descriptive** – include the course information (W20 BIOL4285), and briefly mention the topic (example: "W20 BIOL4285 Sanger sequencing question"). I may be slower to respond to emails without a descriptive subject line.
- Somewhere in the email have **your name and student number**. You can use the name you want to be addressed by; it doesn't have to be the name on your student card.
- If your question was addressed in the syllabus, in class, or on Moodle you may not get a reply. You are still welcome to ask in class, or during drop-in hours.
- Most questions about course material are more difficult to answer over email than in person, and I may direct you to see me instead. Come by drop-in hours instead or make an appointment to go over the concept with me.
- I may post answers to your questions on the discussion forum (Moodle). Other students may have the same question, and everyone can benefit from a classmate or instructor answering it. If I do so all identifying information will be removed.
- **Harassing or abusive, and rude emails will not be tolerated.** You will be reported to the department or Faculty or simply ignored, depending on severity.

There are certain types of email that will never receive a reply.

- **Do not email asking when grades will be posted, you will not get a reply.** Grades for tests and assignments will be posted to Moodle as soon as they are ready.
- **All requests for grade "bumping" will be ignored.** It is your responsibility to earn your grade. Individually adjusting a student's grade without academic merit is unethical and violates the academic integrity of this course.

Forum Code of Conduct

Students are encouraged to participate in the online Moodle Forums to discuss course concepts, organize study groups, and ask questions. Discussions should be polite and respectful, and students are expected to follow these guidelines while using the forums:

- Use a clear, informative subject line. Try to be as specific as possible so that other students and the instructor can respond appropriately.
- Post comments appropriate to the particular discussion. Off-topic posts may be moved or deleted.
- Be respectful! Posts containing personal insults/attacks/intimidation/inappropriate language/profanity will be removed. I will be monitoring for disruptive behaviour and also encourage you to email me immediately if you notice inappropriate behaviour in

the forums. You must follow the York University Student Code of Conduct at all times (<http://www.yorku.ca/oscr/codeofrr.html>).

- While you may engage in debate on biological topics, such discussions should be respectful, and evidence based.

Any posts that appear to violate this code of conduct, and any post at all, may be edited, moved to a hidden forum, or deleted at the discretion of instructors/moderators. If posts contain violations of academic honesty or the York University Student Code of Conduct further action will be taken. If you notice any inappropriate posts, please contact me (Dr. Da Sylva) immediately.

Disclaimer: While Moodle moderators/instructors attempt to remove/edit objectionable/inappropriate material as quickly as possible, it isn't always possible to review every post in a timely manner. All posts made on the forums express the views and opinions of the post's author and the instructor/moderators cannot be held liable.

Note: While we may review posted material, we are often unable to correct wrong answers or incorrect information in a timely fashion. You are responsible for judging the accuracy of the information provided.

University Policies

Academic Honesty and Integrity

York students are required to maintain the highest standards of academic honesty and they are subject to the Senate Policy on Academic Honesty (<http://secretariat-policies.info.yorku.ca/policies/academic-honesty-senate-policy-on/>). The Policy affirms the responsibility of faculty members to foster acceptable standards of academic conduct and of the student to abide by such standards.

There is also an academic integrity website with comprehensive information about academic honesty and how to find resources at York to help improve students' research and writing skills, and cope with University life. Students are expected to review the materials on the Academic Integrity website at - <http://www.yorku.ca/academicintegrity/>

Important Note: Numerous students in Faculty of Science courses have been charged with academic misconduct when materials they uploaded to third party repository sites (e.g. Course Hero, One Class, etc.) were taken and used by unknown students in later offerings of the course. The Faculty's Committee on Examinations and Academic Standards (CEAS) found in these cases that the burden of proof in a charge of aiding and abetting had been met, since the uploading students had chosen to ignore the reasonable likelihood of supporting plagiarism in this manner.

To avoid this risk, students are urged not to upload their work to these sites. Whenever a student submits work obtained through Course Hero or One Class, **the submitting student will be charged with plagiarism and the uploading student will be charged with aiding and abetting.**

Note also that exams, tests, and other assignments are the copyrighted works of the professor assigning them, whether copyright is overtly claimed or not (i.e. whether the © is used or not). Scanning these documents constitutes copying, which is a breach of Canadian copyright law, and the breach is aggravated when scans are shared or uploaded to third party repository sites.

Access/Disability

York University is committed to principles of respect, inclusion and equality of all persons with disabilities across campus. The University provides services for students with disabilities (including physical, medical, learning and psychiatric disabilities) needing accommodation related to teaching and evaluation methods/materials. These services are made available to students in all Faculties and programs at York University.

Student's in need of these services are asked to register with disability services as early as possible to ensure that appropriate academic accommodation can be provided with advance notice. You are encouraged to schedule a time early in the term to meet with each professor to discuss your accommodation needs.

Additional Information:

Counselling & Disability Services - <https://counselling.students.yorku.ca/>
Counselling & Disability Services at Glendon - <https://www.glendon.yorku.ca/counselling/>
York Accessibility Hub - <http://accessibilityhub.info.yorku.ca/>

Ethics Review Process

York students are subject to the York University *Policy for the Ethics Review Process for Research Involving Human Participants*. In particular, students proposing to undertake research involving human participants (e.g., interviewing the director of a company or government agency, having students complete a questionnaire, etc.) are required to submit an *Application for Ethical Approval of Research Involving Human Participants* at least one month before you plan to begin the research. If you are in doubt as to whether this requirement applies to you, contact your Course Director immediately.

Religious Observance Accommodation

York University is committed to respecting the religious beliefs and practices of all members of the community, and making accommodations for observances of special significance to adherents. Should any of the dates specified in this syllabus for an in-class test or examination pose such a conflict for you, contact the Course Director within the first three weeks of class. Similarly, should an assignment to be completed in a lab, practicum placement, workshop, etc., scheduled later in the term pose such a conflict, contact the Course director immediately. Please note that to arrange an alternative date or time for an examination scheduled in the formal examination periods (December and April/May), students must complete an Examination Accommodation Form, which can be obtained from Student Client Services, Student Services Centre or online at http://www.registrar.yorku.ca/pdf/exam_accommodation.pdf (PDF)

Student Conduct in Academic Situations

Students and instructors are expected to maintain a professional relationship characterized by courtesy and mutual respect. Moreover, it is the responsibility of the instructor to maintain an appropriate academic atmosphere in the classroom and other academic settings, and the responsibility of the student to cooperate in that endeavour. Further, the instructor is the best person to decide, in the first instance, whether such an atmosphere is present in the class. The policy and procedures governing disruptive and/or harassing behaviour by students in academic situations is available at - <http://secretariat-policies.info.yorku.ca/policies/disruptive-andor-harassing-behaviour-in-academic-situations-senate-policy/>