

Department of Biology Course Outline

SC/BIOL 4285 3.0 Human Molecular Genetics
Fall 2022-3

Course Description

The main objective of the course is to develop an understanding of basic/molecular genetic and genomic techniques we use in order to study human genetic diseases and human evolution. As part of these techniques, the course will concentrate on the analysis of Mendelian genetic traits, and genomic-related concepts related to human health and disease. We will discuss the implication of the emerged genomics field on basic science and on the future of medical treatment and disease prevention.

Learning Outcomes

Upon completion of the Biology of Cancer course, students will

- be able to explain concepts related to different mechanisms involved with the development of genetic diseases
- be able to explain concepts related to different types of treatments for genetic disease.
- be able to describe the role epigenetics in the development of some diseases.
- develop and use critical thinking skills related to the basic genetics
- use the scientific process and scientific data from this course as a basis for the understanding of other biological systems.

Prerequisite or co-requisite

- SC/BIOL 3130 3.00 or SC/BCHM 3130 3.00.
will be enforced in all cases.

Course Instructors and Contact Information

Course Director: Dr. Motti Anafi:
moanafi@yorku.ca

I will usually be available after each in-class meeting to address individual questions. If you need to speak with me out of class, please send me an email to set an appointment.

Emailing the Course Director

Your email will be read and answered as soon as possible. However, I will open only e-mails that fulfill the following requirements:

- Your email must be sent from your regular yorku.ca email account (**not from the eClass server**). As much as possible, do not use non-yorku.ca accounts (such as Hotmail or personal Gmail). Emails from non-yorku.ca accounts or the eClass will likely languish in a spam folder that is checked only intermittently.
- Be sure to include your full name and student number in your email text.
- Your email must include "BIOL/4285" in the subject line. (I am teaching other courses).
- Your email must NOT include an attachment.

Course Content and optional reading

PART I: The Genetic Revolution-from Traditional Genetics to Synthetic Biology

- EMBO Rep. 2009 August; 10(Suppl 1): S50–S53.
<http://www.ncbi.nlm.nih.gov/pmc/articles/PMC2726003/pdf/embor2009156.pdf>
- EMBO Rep. 2009 August; 10(Suppl 1): S42–S45.
<http://www.ncbi.nlm.nih.gov/pmc/articles/PMC2725998/pdf/embor2009130.pdf>
- Hamilton Smith (2014) Synthetic Biology for Genetic Engineering in the 21st Century
<http://www.mediatheque.lindau-nobel.org/videos/33653/2014-synthetic-biology-for-genetic-engineering-in-the-21st-century/laureate-smith-2>
- *Nature* 456, 310-314 (2008) <http://www.nature.com/news/2008/081119/pdf/456310a.pdf>

PART II: Concepts in Mendelian Genetics.

Every basic genetic textbook can be used as a reference.

PART III: Mendelian Traits in Humans

- <http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=gnd>
- <http://www.ncbi.nlm.nih.gov/omim>
- <http://www.dnafb.org/>
- <http://www.ygyh.org/>
- <http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=cooper&part=A2484>
- <http://www.nlm.nih.gov/medlineplus/geneticsbirthdefects.html>

PART IV: Methods in Genomics, Transcriptomics and Epigenetics.

- *Nature Reviews Genetics* 11, 31-46 (January 2010)
<http://www.nature.com/nrg/journal/v11/n1/abs/nrg2626.html>
- *Hum Genet.* 2011 Apr;129(4):351-70 <http://www.ncbi.nlm.nih.gov/pubmed/21331778>
- *Nature* 461, 814-818 (8 October 2009)
<http://www.nature.com/nature/journal/v461/n7265/abs/nature08390.html>
- *Nature* 415, 530-536 (31 January 2002)
- Cold Spring Harb Protoc 2009. Chromatin Immunoprecipitation (ChIP)
<http://cshprotocols.cshlp.org/content/2009/9/pdb.prot5279.full.pdf+html>
- *BMC Bioinformatics* 2009, 10:80 doi:10.1186/1471-2105-10-80
<http://www.biomedcentral.com/content/pdf/1471-2105-10-80.pdf>
- <http://www.genome.gov/>
- *Current Opinion in Biotechnology* 2000, 11:581–585
http://www.who.edu/science/B/people/mhahn/Broder_581.pdf
- *Nature Reviews Genetics* 9, 356-369 (May 2008)
<http://www.nature.com/nrg/journal/v9/n5/abs/nrg2344.html>

- PLoS ONE 2(12): e1361.
<http://www.plosone.org/article/fetchObject.action?uri=info%3Adoi%2F10.1371%2Fjournal.pone.0001361&representation=PDF>
- <http://www.genome.gov/26525384>

PART V: Ancient Genomics

- Nature 499, 74–78 (04 July 2013)
<http://www.nature.com/nature/journal/v499/n7456/abs/nature12263.html>
- Nature 468, 1053–1060 (23 December 2010)
<http://www.nature.com/nature/journal/v468/n7327/pdf/nature09710.pdf>
- Published online 6 May 2010 | Nature
<http://www.nature.com/news/2010/100506/full/news.2010.225.html>

Disclaimers

The information presented in the lectures is provided for educational purposes only and should not be considered as medical advice.

Evaluation

Mid-Term Exam 1 25%
Date: Friday, Oct 7, 2022
Time: 11:30am
Duration: 60 minutes

This exam will be on **Parts 1 and 2** of the course.

Mid-Term Exam 2 25%
Date: Friday, Nov 18, 2022
Time: 11:30pm
Duration: 60 minutes

This exam is **cumulative**, but it will concentrate on **Parts 2-3** of the course (both basic and human mendelian are connected)

Final Exam 50%
Date: During the formal Fall examination period. Will be published by the registrar's office.
Duration: 150 minutes

All exams are cumulative: everything discussed in the pre-recorded lectures, applications and related principles from the required readings is a “fair game” on the exam.

Mid-terms and final exams will take place in class only. NOT online. You must be in class to write them.

Experiential Education and E-Learning

The Course eClass web site

To access eClass, please follow the instructions below.

1. Go to: <https://eclass.yorku.ca/eclass/my/>
2. Login with your Passport York account.

Here you will find

- An updated course outline with optional reading
- Discussion Forum: here students can discuss the course material, ask questions about the material, etc.
- Announcements
- Grades
- Pre-recorded lectures
- Documentation

Please note that the course director's announcements on the eClass take precedence over any other information (especially if you are communicating with each other via WhatsApp etc.).

If you have technical eClass-related questions, please direct them to UIT Client Services at 416-736-2100 x55800 or email helpdesk@yorku.ca.

Schedule

Course Schedule

Course Schedule and Location: Fridays at 11:30 am in CB 115

Teaching methods

- In the last couple of years, we had at the university many discussions about the best teaching methods for "online" and "in-person" courses. As for BIOL4285 Fall 2022-3, I decided to take the best from both methods and combine them together. I decided to use **here flipped course strategies**.
- **"What is a flipped classroom strategy?** Flipped lessons replace teacher lectures with instructional material—often a prerecorded lecture—that students watch and interact with at home. Later, they apply what they learned at home from the prerecorded lectures in sessions in-class through various activities such as questions answers and discussions."
- Accordingly, the lectures and the discussions are going to be delivered in two different modes: **First, students need to cover the relevant pre-recorded lecture/s on their own, and second, we will have In-class / in-person meetings for further discussions of the material covered in the pre-recorded lectures, including Q & A sessions, after exam reviews and much more.**

- The online portion will be online only (I will not repeat the entire lecture in class)
- The in-class portion will be in-class only. For many reasons, the in-class session will not be recorded. **YOU NEED TO ATTEND THE CLASS FOR IN-CLASS ACTIVITIES.** The “in-person” meetings are not going to be recorded.
- Still, I opened for you an activity called WIKI where students can post summaries of the in-class activities. Furthermore, the FORUM can be used for questions and discussions as well.
- **All exams (mid-terms and final) will take place in class only.** There is no online version for these evaluations.
- The pre-recorded lectures consist of the complete material of the course. As for the in-class portion of the course: We will meet in CB 115 once a week. It is highly recommended for students to attend these meetings.
- The pre-recorded lectures will be posted in three “waves”: the first cluster, the lectures for the chapters to be covered on the first mid-term. Later, as the second cluster, the lectures cover the second mid-term. Later, I will post the rest of the material for the course.
- Students can use the delivery method in quite a flexible way: For example, you can access the visual material covered in high-resolution pre-recorded lectures at any time convenient to you as many times as you wish. You will have the flexibility to view the entire lecture at once or to stop the lecture at any stage of the lecture. You can run the lecture more quickly or slowly. You can turn down/up the audio as you wish.

Few tips on how to study for the course:

- As for the exams, you must know and understand the material presented in the pre-recorded lectures.
- The optional readings can help students consolidate and expand their understanding of the material. However, much of these resources will not be covered in class. On the exams, I will **concentrate** on topics covered in the pre-recorded lectures and their applications. However, reading the optional reading material and attending the “in-class” meetings are likely to be very helpful.
- The material presented in the lectures and other components of the course such as tests and exams have been developed from a large variety of resources, including websites, textbook supplements and other material mentioned.
- I will usually be available after each in-class meeting to address individual questions. If you need to speak with me out of class, please send me an email to set an appointment.

Course Policies

Tests and Exams

- No opportunities to make up missed mid-term exams will be offered. In all cases of missed mid-term exam, the percentage value of the missed mid-term will be added to the final exam.
- If the final exam is missed, the student must petition your home faculty for permission to write the final exam.
- **If the petition will be granted:** The level of difficulty and the material covered on deferred final exam will be similar to the original exam. However, **the format of all deferred mid-terms/final exams is likely to be different from the original exam (e.g., short answer questions or oral exams instead of multiple-choice questions).**
- If the deferred final exam is missed the student must petition their home faculty again for a permission to write a second deferred final exam. **If the petition will be granted the student will be evaluated on an oral exam.**
- No doctor notes or any other documentation are required for missed mid-terms. For the petition for missed final exams or for the petition to write second deferred exams, the documentations needed are according to the policy of your home faculty.
- It is your responsibility to ensure that you are available to sit for final examinations during the entire exam period for the Fall term (Dec 8-23, 2022)

Rules for viewing term tests:

After each exam, we will have an academic feedback session in the following in-class meeting.

If you are interested to view your exam and comparing it against the key, you need to send an e-mail to the course director **by two weeks after the day the grades were posted** on the eClass. Every exam viewing session will be up to 20 minutes for viewing the exam and comparing it against the detailed key. During test viewing sessions the regular examination rules will apply. If after you viewed your exam against the key, you feel that you deserve more marks you can send an e. mail to the course director (Attn: Dr. Motti Anafi, e-mail: moanafi@yorku.ca).

Copyright protection of the posted pre-recorded lectures

- 1) The material presented in the pre-recorded lectures has been developed from a large variety of resources, including websites and textbooks.
- 2) I am doing my best to post the credit for the developers of each external resource that was included in my lectures. However, in some cases, the original material is no longer available on the web, and finding the person or organization that deserved the credit may not be possible despite my efforts.
- 3) The prerecorded lectures are copyright protected by the course director and many third parties, private people, and organizations.

4) The prerecorded lecture will be available for you through the course on the eClass. You can use them in the eClass, but if you wish, **From the eClass** you can go to the source of the lectures on YouTube by clicking the YouTube link on the lower right of each video lecture on the eClass. On YouTube, you will find some closed captions and some other functions. The prerecorded lectures are “unlisted” on YouTube- **do not share the link with others and do not use the link directedly. You need to go to eClass first and sign in with your York Password, and from the eClass, you will be able to go to the source on YouTube if you wish to do so.**

5) Students are NOT allowed to copy the videos and/or to post them elsewhere, directly or as an embedded link.

6) Not complying with any of the above will be considered an infringement of copyright law.

Important Dates

EVENT	FALL (TERM F)	YEAR (TERM Y)	WINTER (TERM W)
Classes start	Sept. 7	Sept. 7	Jan. 9
Last date to announce components of final grades	Sept. 20	Sept. 20	Jan. 23
Fall Reading Week ¹	Oct. 8-14	Oct. 8-14	
Last date to submit Fall term work	Dec. 6	Dec. 6	
Fall classes end	Dec. 6	Dec. 6	
Fall Study Day ²	Dec. 7	Dec. 7	
Fall examinations ³	Dec. 8-23	Dec. 8-23	
Winter Reading Week ¹		Feb. 18-24	Feb. 18-24
Last date to submit Winter term work		April 8	April 8
Winter classes end ⁴		April 10	April 10
Winter Study Days ²		April 11	April 11
Winter examinations ⁵		April 12-27	April 12-27
Notes		Virtual Friday required due to Good Friday: Monday, April 10	Virtual Friday required due to Good Friday: Monday, April 10

For more information go to: <https://registrar.yorku.ca/enrol/dates/2022-2023/fall-winter>

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 A note from the Faculty of Science Committee on Examinations and Academic Standards: 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 been found in all cases to be wilfully blind to the reasonable likelihood of supporting plagiarism in this manner. Accordingly, 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.

Students 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. Please note that registering with disabilities services and discussing your needs with your professors is necessary to avoid any impediment to receiving the necessary academic accommodations to meet your needs.

Additional information is available at the following websites:

Counselling & Disability Services - <http://cds.info.yorku.ca/>

Counselling & Disability Services at Glendon - <https://www.glendon.yorku.ca/counselling/>

York Accessibility Hub - <http://accessibilityhub.info.yorku.ca/>

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 and submit an [Examination Accommodation Form](#) at least 3 weeks before the exam period begins. The form can be obtained from Student Client Services, Student Services Centre or online at

https://registrar.yorku.ca/sites/registrar/files/pdf/exam_accommodation.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/>

More information:

Students in Crisis

Students in crisis should contact the Office of Student Community Relations (OSCR) at 416 736-5231 or oscr@yorku.ca .

Website: <https://oscr.students.yorku.ca>

Students not in Crisis but Needing Support/Help

Below are some links to resources for students who need support but are not necessarily in crisis.

1. **Academic Advising** - Each Faculty has its own Academic Advising Office

Science: <https://www.yorku.ca/science/students/current-students/academic-advising/>

Health: <https://www.yorku.ca/health/academic-advising/>

LAPS: <https://www.yorku.ca/laps/support/academic-advising/>

2. **ESL-** Support for ESL (English as a Second Language) Students URL

3. **Study/Learning Skills** - Learning Skills Office (Learning Skills Workshops etc) : <https://www.yorku.ca/scll/learning-skills/>

4. Student Counselling and Development:

<https://counselling.students.yorku.ca>

phone: 416-736-5297

After Hours Support

If students need assistance outside of SCD's regular operating hours, they can call the following counselling/crisis/support lines:

- Local emergency personnel (fire, ambulance, police): 911
- [York Security Services](#) (emergency): 416-736-5333
- [York Security Services](#) (non-emergency): 416-650-8000
- [goSAFE](#): 416-736-5454
- [Good2Talk](#): Call 1-866-925-5454 or text GOOD2TALKON to 686868
- [Assaulted Women's Helpline](#): 416-863-0511
- [Burlington Distress Centre](#): 905-681-1488
- [Distress Centers of Greater Toronto](#): 416-408-HELP (4357)
- [Durham Crisis and Mental Health Line](#): 905-666-0483
- [Gerstein Centre Crisis Line](#): 416-929-5200
- [Oakville Distress Centre](#): 905-849-4541
- [Scarborough Health Network Community Crisis Line](#): 416-495-2891
- [Spectra Distress Line](#): 905-459-7777
- [York Community Crisis Response Services](#): **1-855-310-COPE (2673)**
- [24/7 Crisis Support Peel Dufferin](#): 905-278-9036
- [211Ontario](#): 211