

BIOL 5591

Advanced Genomics

Spring 2025

MR 5:15 - 6:55 pm | BEHR 030

Professor: Jamie Henzy

Office: Mugar 205F

Office hours: by appt

Material: Readings consist of a mix of research papers and reviews available through PubMed; some of the exercises require online genomics tools that you can access on a laptop.

Prerequisite: BIOL 2301 Genetics and Molecular Biology

Course objectives: Generally, become familiar with the approaches, techniques, and tools of analysis that genomicists use and the types of research questions they pursue, for example:

- Describe various techniques used in sequencing, assembling, and annotating genomes.
- Explain techniques used to determine, analyze, and visualize transcriptional information.
- Demonstrate understanding of the concepts underlying phylogenetic analysis.
- Interpret data and findings from research articles.
- Describe how structure prediction is performed.

Course format: Please note that this is a fully in-person course. Class sessions will include a mix of lecture, discussion of papers, and exercises. Twice during the semester pairs of students will give presentations, followed by a class quiz.

Grading:	Paper summaries:	15 pts x 9	135
	Quiz+discussions:	(5 + 15) pts x 9 (drop lowest)	160
	Exercises:	15 pts x 10	150
	Presentation:	50 pts	50
	TBD	5 pts	5
	In-class exams:	250 pts x 2	500
			1000

Grading scale:

A: 90 - 100% B: 80 - 89% C: 70 - 79% F: 69% or less

Pluses and minuses will distinguish between higher or lower performance within each letter range. To ensure fairness, everyone will be graded according to the same standards. If you identify a calculation error in any score, please let me know. However, please do not ask for special consideration in terms of extra credit, more generous rounding, or any other type of "grade bump". Such requests are inappropriate, unprofessional, and have zero chance of being granted.

Paper summaries: A major component of the course involves reading and summarizing papers, of which there are nine. Some papers will be research articles and others, reviews. In each case, you'll have a template to guide your summary, and you'll be quizzed in class on the contents.

Quiz+discussions: For each paper, there will be a 5-pt in-class quiz on the day we discuss the paper. The quiz will come directly from components of the summary template, such as the major goal of the paper and key results.

After the quiz we'll discuss the paper. Provided you submitted the summary by the start of class, you earn 15 discussion points, even if I don't call on you to answer a question. The discussion pts are only available if you have submitted your summary **by the start of class**. Since absence on a discussion day (see schedule below) will cost 5 pts, as a buffer against the various events (sickness, travel, "mental

health" days) that might prevent you from attending class on any of these dates, one discussion score will be dropped.

Exercises: There will be 10 exercises worth 15 pts each to introduce you to various genomics methods and techniques. You'll have a week to complete each, after which solutions will be posted. **No submissions will be accepted after posting of the solutions.**

Presentation: Pairs of students will each give a 5-min presentation on selected topics from the papers we have discussed. Presentations will take place at the start of classes on Thursdays, with no more than two pairs presenting. See the signup sheet to choose a date.

Exams: There will be two in-class exams.

Exam 1: papers 1 – 4, exercises 1 – 4, lecture slides

Exam 2: papers 5 – 8, exercises 5 – 8, lecture slides

Getting help: Please post any questions about course material or assignments -- questions that other students may also be interested in -- in the "Ask the prof" Discussion forum on Canvas. For more specific issues, please sign up for a Zoom office hours slot, using the link found on the Office Hours page.

Expanding the field: Although science is considered an "objective" endeavor, most research questions, discoveries, and applications have been influenced by the experiences of the scientists involved, who have been predominately white males. Science and the society it serves is greatly enriched by the participation of people from diverse backgrounds, who can bring fresh views and priorities, driving new lines of inquiry and application. A hurdle is that students who have traditionally been under-represented in science often feel insecure in their ability to participate and express their views. A secret: anyone who has curiosity and wants to use data to solve a mystery is a scientist at heart! Please let me know if you have any concerns of this nature.

Cheating policy: Honesty, integrity, and ethical behavior are of utmost importance in science, and can make or break a scientific or medical career. Anyone caught cheating (for example, copying from the work of other students, or collaborating during quizzes) will receive a zero and be referred to the Office of Student Conduct and Conflict Resolution. Please make sure you have read and understand the student code of conduct: <http://www.northeastern.edu/osccr/code-of-student-conduct/>

Schedule of Topics: Following the schedule is the list of readings. All readings are freely accessible through the Northeastern PubMed database or elsewhere online.

Date	Topic	Exer.
Mon Jan 6	Introduction to Genomics	
Thu Jan 9	Genome sequencing	
Mon Jan 13	Discussion: Burian et al.	
Thu Jan 16	SARS-CoV2 genome	1
Mon Jan 20	Holiday	
Thu Jan 23	Discussion: Lu et al.	2
Mon Jan 27	Genome of Borrelia, agent of Lyme disease	
Thu Jan 30	Discussion: Chaconas et al.	3
Mon Feb 3	Ancient DNA	
Thu Feb 6	Discussion: Klunk et al.	4

Mon Feb 10	Spillover and loose ends	
Thu Feb 13	Exam 1	
Mon Feb 17	Holiday	
Thu Feb 20	Transcriptomics and RNA-seq	5
Mon Feb 24	Discussion: Uffelmann et al.	
Thu Feb 27	Transposable elements	6
Mon Mar 3	Spring break	
Thu Mar 6	Spring break	
Mon Mar 10	Discussion: Du et al.	
Thu Mar 13	Regulatory elements	7
Mon Mar 17	Discussion: Mazo-Vargas et al.	
Thu Mar 20	Linkage and genome-wide association studies	8
Mon Mar 24	Discussion: Horowitz et al.	
Thu Mar 27	Spillover and loose ends	9
Mon Mar 31	Exam 2	
Thu Apr 3	TBD	
Mon Apr 7	Discussion: TBD	
Thu Apr 10	TBD	10
Mon Apr 14	TBD	

Reading list *amenable to changes based on student interest or publication of new research papers:*

1. **Burian AN et al.** Genome sequencing guide: An introductory toolbox to whole-genome analysis methods. *Biochem Mol Biol Educ.* 2021 Sep;49(5):815-825. doi: 10.1002/bmb.21561. Epub 2021 Aug 11. PMID: 34378845; PMCID: PMC9291972.
2. **Lu R et al.** Genomic Characterisation and Epidemiology of 2019 Novel Coronavirus: Implications for Virus Origins and Receptor Binding. *Lancet.* 2020 Feb 22;395(10224):565-574. doi: 10.1016/S0140-6736(20)30251-8.
3. **Chaconas G, Castellanos M, Verhey TB.** Changing of the guard: How the Lyme disease spirochete subverts the host immune response. *J Biol Chem.* 2020 Jan 10;295(2):301-313. doi: 10.1074/jbc.REV119.008583. Epub 2019 Nov 21. PMID: 31753921; PMCID: PMC6956529.
4. **Klunk J et al.** Evolution of Immune Genes Is Associated with the Black Death. *Nature.* 2022 Nov;611(7935):312-319. doi: 10.1038/s41586-022-05349-x.
5. **Uffelmann E et al.** Genome-wide association studies. *Nat Rev Methods Primers.* 2021;1:59. doi: 10.1038/s43586-021-00056-9.
6. **Du AY et al.** Regulatory transposable elements in the encyclopedia of DNA elements. *Nat Commun.* 2024 Jan 8;15:7594. doi: 10.1038/s41467-024-51921-6.
7. **Mazo-Vargas A et al.** Deep cis-regulatory homology of the butterfly wing pattern ground plan. *Science.* 2022 Oct 21;378(6619):304-308. doi: 10.1126/science.abi9407.
8. **Horowitz JE, Kosmicki JA, Damask A et al.** Genome-wide analysis provides genetic evidence that ACE2 influences COVID-19 risk and yields risk scores associated with severe disease. *Nat Genet.* 2022 Mar;54(3):382-392. doi: 10.1038/s41588-021-01006-7.
9. **Li L et al.** Recurrent gene flow between Neanderthals and modern humans over the past 200,000 years. *Science.* 2024 Jul 12;385(6705):ead1768. doi: 10.1126/science.adi1768. Epub 2024 Jul 12. PMID: 38991054.

OR

9. Hughes JF et al. A gene deriving from the ancestral sex chromosomes was lost from the X and retained on the Y chromosome in eutherian mammals. *BMC Biology* (2022) 20:133. <https://doi.org/10.1186/s12915-022-01338-8>. **OR**

9. Dopkins N et al. Ribosomal profiling of human endogenous retroviruses in healthy tissues. *BMC Genomics*. 2024 Jan 2;25(1):5. doi: 10.1186/s12864-023-09909-x. PMID: 38166631; PMCID: PMC10759522.