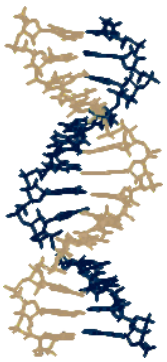
BIOS 4530/BIOL 8530 - Human Evolutionary Genomics

Fall 2022 Syllabus

Fall 2022 semester, 3 credits
Monday and Wednesday – 3:30pm-4:45pm
Location: Howey Physics N210



Instructor

Dr. **Joseph Lachance**
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Office: 2103 Krone Engineered Biosystems Building
Office hours: after each class or by appointment

Overview

This is an advanced course where students will discuss primary literature and use computational tools to investigate how evolution has shaped global patterns of human genetic variation. It is geared towards both undergraduate students and graduate students (MS and PhD). This class integrates genetics, evolutionary biology, anthropology, computational biology, and bioinformatics. Each week students will be introduced to:

- ***concepts*** of evolutionary genetics
- ***papers*** of cutting-edge research in human genomics
- ***exercises*** where computational tools are applied to real-world datasets

Prerequisites

Undergraduates taking BIOS 4530 are required to have previously taken an introductory course in undergraduate biology (or tested out due to AP credit). Previous coursework in genetics or evolutionary biology is beneficial, but not required. It is assumed that you are comfortable using a command line interface.

Coronavirus contingencies

The health of everyone in our class is my primary concern. If you happen to get COVID or another illness during the fall semester, please let me know so that accommodations can be made.

Travel/homework days

I will be attending an African cancer genetics meeting in late September and the annual meeting of the American Society of Human Genetics in late October. Because of this, a small number of our classes will be asynchronous (check the schedule for specific days). Bonus recordings of additional class material may end up being posted via Canvas during these travel days. You are encouraged to stop by Howey Physics N210 at our regular class time during "homework days" which feature an active learning exercise.

Learning outcomes

As a result of taking this course, you will be able to:

1. Understand how different evolutionary forces shape human genomic variation. This will enable you to propose alternative hypotheses to explain observed patterns of human genetic diversity.
2. Critically read and discuss the scientific literature. You will be able understand how a given paper advances our knowledge of human evolutionary genomics and be able to critically evaluate both the approaches taken and the conclusions of the authors.
3. Use computational tools to analyze genome-scale datasets of human genetic variation. You will become comfortable working with large datasets and develop a familiarity with a wide array of computational tools. This hands-on experience will serve as a springboard for your own research.

Textbook and class webpage

Optional textbook: *Human Evolutionary Genetics (2nd edition)* by Jobling et al. (2013).
Optional companion text: *The Brief History of Everyone Who Ever Lived* by Rutherford (2016)
Additional online resources, including .pdfs of papers: http://canvas.gatech.edu

Software

RStudio: https://www.rstudio.com/products/rstudio/download/
SLiM for macOS or Linux can be downloaded from: https://messerlab.org/slim/
PLINK: https://www.cog-genomics.org/plink/

Instructional format

Classes involve a combination of lectures, critical evaluation of the primary literature, and active learning exercises.

Lectures and journal club discussion: Each week our class will focus on a different topic. Traditional lectures will be used to help ensure you have foundational knowledge about this topic. Each week we will also discuss two scientific papers (.pdfs of which are available via Canvas). Read the abstracts of both papers, choosing one paper to skim and one paper to read carefully. Write at least one discussion question related to these papers. To receive credit, upload your questions to Canvas. Warning: I will call on random people in class, so please read the papers!

Active learning exercises: In this class you'll have a chance to play with real genomic data, explore online databases, and get a taste of some of some computational tools that are available to study human evolution. Each week we will feature an active learning exercise that complements what you have learned in lecture and read about in the literature. Note that these active learning exercises require a computer, so bring a laptop or work through the worksheet with a neighbor in class. Those students who already have advanced expertise are encouraged to help their classmates. Each worksheet will also include stretch goals. To receive credit, upload the relevant worksheet for each active learning exercise to Canvas

Course policies and assignments

Participation: You are expected participate in both the discussion of the primary literature (journal club) and in the weekly active learning exercises. Please also see http://www.catalog.gatech.edu/rules/4/ for more information about institute expectations and restrictions around attendance, including information about excused absences. Participation in journal club discussions and active learning exercises will be assessed using a check/no-check scale. If you are unable to attend class due to illness or another type of excused absence, please email me beforehand.

Take home exam: There will be one take-home exam in this course. Think of this exam as a rigorous homework assignment. A .pdf version of your completed exam will need to be uploaded to http://canvas.gatech.edu. Students are allowed to research their answers over the internet. However, take-home exams must represent the work of individual students. In other words, make sure that your answers are your own. Ignoring this policy will result in a violation of the Georgia Tech Honor Code and a score of zero for the exam. Late take-home exams will be accepted up to 2 days late, with 20% deducted per 24-hour period that it is late.

Digest article: Undergraduate students in BIOS 4530 will write a short (~600 word) summary of a research article. The goal of this assignment is to assess your ability to comprehend the primary literature. Note that the article you pick must not be a review paper, and it must cover a topic in human evolutionary genomics. Please check with the course instructor early on to discuss the topic and scope of this paper. Additional details of what goes into a Digest can be found at: https://sites.duke.edu/evodigests/writing-instructions/. You will have a chance to get feedback from your classmates a couple weeks before the deadline of this assignment.

Specific Aims page: Graduate students in BIOL 8530 will write a Specific Aims page on a research topic in human evolutionary genomics. Ideally this short fellowship proposal will overlap with your MS or PhD research. That said, it doesn't have to be something you will actually do! This one-page summary must outline a clear research question, explain why this question is important, contain a testable hypothesis, and include two or three specific aims which outline how you will attack the research question. Please check with the course instructor early on to discuss your research topic. You will have a chance to get feedback from your classmates a couple weeks before the deadline of this assignment.

Graduate and undergraduate expectations

A higher standard of rigor is required for graduate students taking BIOL 8530, as opposed to undergraduates taking BIOS 4530. Grading will reflect these differences. Graduate students taking BIOL 8530 are expected to take a more active role in our discussions of the primary literature than students taking BIOS 4530. Active learning exercises will be graded the same for both sets of students (on a check/no-check scale). One key difference between the two sections of this course is that undergraduates taking BIOS 4530 will write a Digest article, and graduate students taking BIOL 8530 will write a Specific Aims page.

Evaluation

Overall grades in this class will be curved, with the following breakdown:

Journal club questions	15%
Active learning exercises	15%
Take-home exam (due October 14)	35%
Digest article/Specific Aims page (due December 9)	35%

Grades will be curved (i.e., we will not use a 90-80-70 scale). Historically, over two-thirds of students who have taken this course received an A.

Academic integrity

There is a zero-tolerance regarding cheating and plagiarism. If you have any questions about what constitutes plagiarism, please email the course instructor. Students are reminded of the obligations and expectations associated with the Georgia Tech Academic Honor Code: http://policylibrary.gatech.edu/student-affairs/academic-honor-code. Violations of the honor code can result in a student receiving an "F" in this class.

Student-Faculty Expectations Agreement

At Georgia Tech we believe that it is important to strive for an atmosphere of mutual respect, acknowledgement, and responsibility between faculty members and the student body. See http://www.catalog.gatech.edu/rules/22/ for an articulation of some basic expectation that you can have of us and that we have of you. In the end, simple respect for knowledge, hard work, and cordial interactions will help build the environment we seek. Therefore, we encourage you to remain committed to the ideals of Georgia Tech while in this class.

Additional campus resources

- Center for academic success: http://success.gatech.edu/
- Communication center: http://www.communicationcenter.gatech.edu
- Counseling Center: http://counseling.gatech.edu; 404-894-2575

Learning accommodations

If needed, accommodations will be made for students with disabilities. These accommodations must be arranged in advance and in accordance with the Office of Disability Services.

Statement of intent for inclusivity

As members of the Georgia Tech community, we are committed to creating a learning environment in which all of our students feel safe and included. Because we are individuals with varying needs, we are reliant on your feedback to achieve this goal. To that end, we invite you to enter into a dialogue with us about the things we can stop, start, and continue doing to make our classroom an environment in which every student feels valued and can engage actively in our learning community.

Schedule for Human Evolutionary Genomics (Fall 2022)

Data	Topic	Readings / active learning exercise
Aug 22 (Mon)	Variation and ascertainment bias	1000 Genomes Project Consortium 2015
Aug 24 (Wed)		**UCSC genome browser + dbSNP**
Aug 29 (Mon)	Population genetics theory	Kern and Hahn 2018; Jensen et al. 2018
Aug 31 (Wed)		**RedLynx + R: allele frequencies**
Sep 5 (Mon)	No class (Labor Day)	
Sep 7 (Wed)	Linkage and multilocus genetics	Machiela and Chanock 2015; Peñalba and Wolf 2020
Sep 12 (Mon)		**LDlink**
Sep 14 (Wed)	Geography and genetics	Peter et al. 2020; Currat et al. 2021
Sep 19 (Mon)	No class (homework day)	**R: PCA + GGV Browser**
Sep 21 (Wed)	No class	
Sep 26 (Mon)	Out-of-Africa migration	Scerri et al. 2019; Domínguez-Andrés et al. 2019
Sep 28 (Wed)		**SLiM: simulating founder effects**
Oct 3 (Mon)	Inferring demographic history	Schlebusch et al. 2020; Font-Porterias et al. 2021
Oct 5 (Wed)		**SLiM: simulating gene flow**
Oct 10 (Mon)	Ancient introgression	Skov et al. 2020; Gower et al 2021
Oct 12 (Wed)		**R: ABBA/BABA tests**
Due: Oct 14	**Take-home exam**	**(assigned Oct 7)**
Oct 17 (Mon)	No class (fall break)	
Oct 19 (Wed)	Human adaptation	Sugden et al. 2018; Rees et al. 2020
Oct 24 (Mon)	No class (homework day)	**R: PBS statistics + GGV Browser**
Oct 26 (Wed)	No class	
Oct 31(Mon)	Genetic load	Lynch 2016; Roth et al. 2016
Nov 2 (Wed)		**R: quantifying deleterious alleles**
Nov 7 (Mon)	Evolutionary medicine	Crespi 2020; Benton et al. 2021
Nov 9 (Wed)		**aDNA exercise (to be decided)**
Nov 14 (Mon)	GWAS and evolution	Adhikari et al. 2019; Privé et al. 2022
Nov 16 (Wed)		**NHGRI-EBI GWAS Catalog + PLINK tutorial**
Nov 21 (Mon)		**Peer feedback re: Digests/Specific Aims**
Nov 23 (Wed)	No class (student recess)	
Nov 28 (Mon)	Evolution of polygenic risk scores	Rosenberg et al. 2019; Durvasula et al. 2021
Nov 30 (Wed)		**R: polygenetic risk scores**
Dec 5 (Mon)	The future of human evolution	
Due: Dec 9	**Digests/Specific Aims**	

All details in this syllabus are subject to change!