

BIOI 397/495 / BIOL 395 Bioinformatics Survey/Special Topics – Human Genetics

- Instructor: Dr. Heather Wheeler hwheeler1@luc.edu
- Prerequisites: BIOL 282 or Bioinformatics MS student
- Days/Times: Tues/Thurs 2:30 – 3:45PM
- Classroom: IC 111 (starting Feb 1) or Zoom (Jan 18-27, see Zoom Pro tab on Sakai for link)
- Office Hours: Mondays 4:00-5:00PM, LSB 324 or Zoom (see Zoom Pro tab on Sakai for link)

Objectives:

- Understand the foundations of human complex trait genetics
- Critically analyze and discuss primary literature in human genetics
- Increase proficiency in scripting, command line tools, and visualizations used in the field
- Design, implement, and evaluate a genome-wide analysis of human phenotype(s)

Course Materials: Required reading: *How to Argue With a Racist: What Our Genes Do (and Don't) Say About Human Difference* by Adam Rutherford (ISBN: 978-1615198306). We will also use free online tools, open source software, and the scientific literature. Students must have access to a laptop (Mac, Windows, or Linux operating system) both during and outside of class time with administrative rights, i.e., can install software. Computers must have a minimum of 8 GB of RAM and a reliable internet connection. Students will be given an account to access a remote Linux server for completing homework and the final project. For synchronous Zoom sessions, each student needs a device with a microphone and a large enough screen to view shared code. A webcam is also helpful for interacting with classmates and instructor.

Office Hours & Seeking Help: To meet at a time other than scheduled office hours, you must email the instructor to schedule a day and time in advance. Another, usually faster, route to get assistance is to ask questions via email. I do my best to respond to emails within 24 hours. There are many online forums where you can post questions or search for questions and answers. When you encounter an error message, you can always try and **Google it!**

Student Accommodations: Any student requesting accommodations is required to register with Student Accessibility Center (SAC). Students will provide the instructor with an accommodation notification from SAC, preferably within the first two weeks of class. Students are encouraged to meet with the instructor individually in order to discuss their accommodations. All information will remain confidential. For more information or further assistance, please call 773.508.3700 or visit <https://www.luc.edu/sac/>.

Academic Integrity: Plagiarism and cheating will not be tolerated and will be reported to the college. See http://www.luc.edu/academics/catalog/undergrad/reg_academicintegrity.shtml.

The homework assignments will be done independently (although you can get help from each other in class). Cheating includes submitting as your own work something that has been written by another person (classmate, tutor, website, etc.). When discussing others' work, simply changing a word or two in a sentence is plagiarism. If you are concerned that your text may border on plagiarism, ask. **If you are found to present someone else's work as your own, you will receive zero points for the assignment. If you present someone else's work as your own a second time, you will receive an F for the course.** Some assignments, including journal clubs and/or the research project, may be completed with a partner. As part of a team, you are expected to contribute to the same degree as your partner. **"Coasting" or letting your**

partner carry you is plagiarism; you are taking credit for contributions that you did not make. This is on-par with presenting someone else's idea/work as your own without proper citation. Plagiarism and cheating of any form will not be tolerated. **As a contributing member of a team, it is your responsibility to ensure that all members are contributing.** Issues that cannot be resolved within the group should be reported to the instructor.

Grading: Your grade will comprise several assignments as listed below.

Assignment	Points
Homework 1-5	25
Journal Club Presentation	10
Journal Club Participation	9
Book Club Participation	10
Book Reflection Paper	9
Research Project Plan	10
Research Project Progress Presentation	10
Research Project Progress Participation	2
Final Research Project Code	10
Final Research Project Paper	15

- **Homework: 25 pts** (5 pts each). Homework assignments will be exercises using human genetics software and web resources with example data. These exercises will introduce you to tools and analyses that you will incorporate into your own Research Project. Homeworks will be posted and turned in to Sakai.
- **Journal Club Presentation: 10 pts.** You will present on one Journal Club date. Planned journal articles and dates are listed in the course syllabus. **Choose one topic you would like to present and sign up here, first come, first served:** https://doodle.com/poll/vgkahi3fug424rvm?utm_source=poll&utm_medium=link. Note, there are 9 Journal Club dates, so you will likely split presenting duties with a partner on your Journal Club date. The Journal Club led by Dr. Wheeler on January 27 will serve as an example presentation. Your presentation will briefly describe the question the paper is trying to answer. This will require presenting background information about the topic that may not be included in the paper itself, i.e., how would you introduce the research topic if you were teaching a class about it? As the presenter(s), you will then engage your classmates in a discussion of the paper. This will likely include asking classmates to describe a figure or table, posing thought questions about the approach and its limitations, answering questions posed by the group or directing them to others, etc. This discussion is expected to last 45-60 min. A rubric of expectations is posted on Sakai Assignments. Your presentation slides (pptx/pdf) must be submitted via Sakai Assignments prior to the scheduled presentation.
- **Journal Club Participation: 9 pts.** When someone other than yourself is the journal club presenter, you are expected to read the presented paper(s) before class and actively participate in the discussion of the paper during class. If you make at least one substantive comment (e.g., describe a figure or table, explain a method or result, answer a thought question posed by the presenter, ask an insightful question, etc.), you will be awarded 1 pt. If you feel you did not get a chance to participate enough during class time, you can continue the discussion on Sakai Discussions by posting a response that was not discussed or a new insightful question about the paper presented to earn your full 1 pt. Discussion posts for a particular journal club must be made before the next class period. Start a new conversation with the date of the journal club if you are the first

- to post to Sakai Discussions for a particular journal club.
- **Book Club Participation: 10 pts.** On Book Club days, Dr. Wheeler will lead the discussion of the assigned pages from the Rutherford book. If you make at least one substantive comment (e.g., describe your reaction to a passage, explain a finding presented in the book, ask an insightful question, etc.), you will be awarded 2 pts. To encourage lively discussion, book club participation points will only be awarded if you speak up in class.
 - **Book Reflection Paper: 9 pts.** Each student will write a 1-2 page informal reflection after reading and discussing the Rutherford book. Discussion prompts and a grading rubric will be posted to Sakai.
 - **Final Project:** You will evaluate the genetics of a complex trait of interest for which GWAS summary statistics are publicly available. You may work independently or with a partner (graduate students must work independently). Expectations will be discussed on Feb 24.
 - **Research Project Plan: 10 pts.** You will outline your project plan according to a template provided in Sakai. Planning ensures your proposed project is feasible and allows you to get feedback from the beginning of your project.
 - **Research Project Progress Presentation: 10 pts.** This presentation will be a 10-minute talk with 3 minutes for Q&A. The presentations should introduce the complex trait, your methods, your results thus far, any hurdles you have encountered, and plan for completion. A grading rubric will be posted on Sakai. The presentation slides (pptx/pdf) must be submitted via Sakai prior to the scheduled presentation.
 - **Research Project Progress Participation: 2 pts.** You can earn up to 2 points for asking questions of other student presentations (1 pt/question). Questions should focus on interpretation of the results or aspects of the methods (rather than simply asking a presenter to repeat/reshow a slide.) Note, time is limited for Q&A. Everyone will not have an opportunity to ask questions of each presentation.
 - **Final Project Code: 10 pts.** You will turn in commented code showing how all the results in your final paper were generated. This will require writing an R markdown document that shows all code used to generate the results included in your final paper. Your homework will give you experience working with R markdown.
 - **Final Project Paper: 15 pts.** Final project topics and plans will be determined by March 15. Your analyses will be reported through a manuscript including the following sections (a grading rubric will be posted on Sakai):
 - Abstract.** 200-250 word summary of the paper.
 - Introduction.** Introduce the complex trait examined. Briefly describe what is known about the genetics of the trait (e.g., Have heritability studies been performed? What did they find? What have prior GWAS found?) Make a connection to what your study does that is new and different while introducing what results are to come in the rest of your paper.
 - Methods.** Include details about the data used, citing where the summary statistics came from. Include descriptions of your analyses and cite computational tools used. Remember, a methods section should provide enough detail that, together with your code, the study can be reproduced.
 - Results.** Describe the results of your investigation. Include figures and/or tables as appropriate.
 - Discussion.** Provide a synthesis of your results and how they fit in with previous studies. Include recommendations for what should be done next. Discuss limitations of your analysis.
 - References.** References cited in the text must be listed. References should be formatted using the PLOS Style (<http://journals.plos.org/plosone/s/submission-guidelines#loc-references>).

110 points are available. Your final grade will simply be the sum of all of the points you have earned over the course of the semester. The minimum points (pts) needed for each grade are listed in the table below. Points are rounded to the nearest tenth.

A ≥ 93 pts	B+ ≥ 87 pts	B- ≥ 80 pts	C ≥ 73 pts	D+ ≥ 67 pts	F < 63 pts
A- ≥ 90 pts	B ≥ 83 pts	C+ ≥ 77 pts	C- ≥ 70 pts	D ≥ 63 pts	

Late/Missed Assignment Policy: Due dates and times for each assignment will be posted on Sakai. LATE ASSIGNMENTS ARE TYPICALLY NOT ACCEPTED. **Extra points are built into the course for this reason.** Exceptions may be made at the discretion of the instructor.

Additional Requirements for Students Enrolled in BIOI 495: Students enrolled in the graduate level course will be required to perform their Research Project independently and include additional analyses to receive full credit for the Final Project as outlined in the rubric presented in class Feb 24.

Class Conduct: One important aspect of your education is learning to respect the rights of others. Please respect others by (1) allowing all classmates the right to voice their questions or opinions without fear of ridicule and (2) not making objectionable (e.g., racist, sexist) comments. Some of the topics we will discuss in human genetics are inherently personal (e.g., ancestry, race, ethnicity). Grant leniency as we try to articulate our thoughts and may stumble over vocabulary. We converse to understand and engage with voices different from our own.

Diversity, Equity, and Inclusion Statement: Science strives to be objective. However, science is historically built on a small subset of privileged voices and thus includes both explicit and implicit biases. My goal is that students from all backgrounds and perspectives are well served by this course. The diversity students bring to human genetics should be viewed as a resource, strength, and benefit as we pursue knowledge in the service of humanity. I intend to present materials and activities that are respectful of diversity: gender, sexuality, disability, age, socioeconomic status, ethnicity, race, religion or no religion, and culture. Your suggestions for improving inclusion are encouraged and appreciated.

Privacy Statement: Assuring privacy among faculty and students engaged in online instructional activities helps promote open and robust conversations and mitigates concerns that comments made within the context of the class will be shared beyond the classroom. As such, recordings of instructional activities occurring during synchronous Zoom sessions will be used solely for internal class purposes by the faculty member and students registered for the course, and only during the period in which the course is offered.

Intellectual Property: All lectures, videos, PowerPoints, and other instructional materials in this course are the intellectual property of the instructor. As a result, they may not be distributed or shared in any manner, either on paper or virtually without the instructor's written permission. Lectures may not be recorded without the instructor's written consent; all lectures will be recorded and available for students of this course only to view in Panopto or Zoom (linked and/or embedded in Sakai) after class. Recognizing that your work, too, is your intellectual property, the instructor will not share or distribute your work in any form without your written permission.

Statement of Intent: By remaining in this course, students are agreeing to accept this syllabus as a contract and to abide by the guidelines outlined in the document.

Spring 2022

Course Schedule

Week	Date	Topics	Pre-class Readings (PDFs of journal articles are on Sakai Resources)	DUE at start of class
1	18-Jan	Course Introduction, Genetics Review		
	20-Jan	Unix Command Line and SSH		
2	25-Jan	Evolution, LD, and GWAS		Homework 1 (Unix)
	27-Jan	<u>Journal Club</u> : Population Structure	Rosenberg et al. 2002 https://doi.org/10.1126/science.1078311 Novembre et al. 2008 https://dx.doi.org/10.1038%2Fnature07331	
3	1-Feb	R tutorial – Principal Component Analysis		
	3-Feb	<u>Journal Club</u> : GWAS Tutorial	Marees et al. 2017 https://doi.org/10.1002/mpr.1608	
4	8-Feb	Heritability		Homework 2 (PCA)
	10-Feb	<u>Journal Club</u> : Phenome-wide Heritability Analysis	Ge et al. 2017 https://doi.org/10.1371/journal.pgen.1006711	
5	15-Feb	Fine mapping tutorial – susieR		Homework 3 (GWAS)
	17-Feb	<u>Journal Club</u> : Integrating Transcriptomics with PrediXcan	Gamazon, Wheeler, Shah, et al. 2015 https://doi.org/10.1038/ng.3367	
6	22-Feb	<u>Journal Club</u> : Polygenic Scores in Disease Risk	Khera, Chaffin, et al. 2018 https://doi.org/10.1038/s41588-018-0183-z	Homework 4 (LocusZoom, susieR)
	24-Feb	Discuss Research Projects, Start Project Plan		
7	1-Mar	Book Club: Introduction	Rutherford Introduction (p. ix-34)	Homework 5 (LD Score, S-PrediXcan)
	3-Mar	<u>Journal Club</u> : Lactase persistence in Africa and Europe	Tishkoff, Reed, et al. 2007 https://doi.org/10.1038/ng1946	
8	8-Mar	No class: Spring Break		
	10-Mar	No class: Spring Break		
9	15-Mar	Book Club: Skin in the Game	Rutherford Part 1 (p. 35-78)	Project Plan
	17-Mar	<u>Journal Club</u> : Complex genetic architecture of skin pigmentation	Martin et al. 2017 https://doi.org/10.1016/j.cell.2017.11.015	
10	22-Mar	Book Club: Your Ancestors Are My Ancestors	Rutherford Part 2 (p. 79-127)	
	24-Mar	<u>Journal Club</u> : Upper Paleolithic humans had recent Neanderthal ancestry	Hajdinjak et al. 2021 https://doi.org/10.1038/s41586-021-03335-3	
11	29-Mar	Book Club: Black Power	Rutherford Part 3 (p. 128-152)	
	31-Mar	<u>Journal Club</u> : GWAS of endurance athlete status	Al-Khelaifi et al. 2020 https://doi.org/10.3389/fgene.2020.00595	
12	5-Apr	Book Club: White Matter	Rutherford Part 4 (p.153-202)	
	7-Apr	<u>Journal Club</u> : GWAS of IQ	Coleman et al. 2019 https://doi.org/10.1038/s41380-018-0040-6	
13	12-Apr	Project Work/Help Day		Book Reflection
	14-Apr	Project Progress Presentations		
14	19-Apr	Project Progress Presentations		
	21-Apr	Project Progress Presentations		
15	26-Apr	Project Progress Presentations		
	28-Apr	Project Work/Help Day		
Finals	6-May			Final Project code, Final Paper

Schedule is subject to change at discretion of the instructor; changes will be published to Sakai.

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