

<https://content.csbs.utah.edu/~rogers/tch/ant5221/index.php>

Human Evolutionary Genetics

Description This course covers theories and methods of molecular population genetics, with emphasis on human examples. Using these tools, genetic data can inform us about population history and adaptive evolution. Laboratory exercises with the Python programming language connect theory to data. Satisfies Quantitative Intensive Requirement.

We will meet twice a week for lecture and once a week for lab. Lectures are in person. For labs, students will log onto Zoom sessions via Canvas.

Prerequisites You should be comfortable with algebra and first-semester calculus. No prior knowledge of Python is needed.

Grading is based on labs, homework assignments, and quizzes. Each lab, homework assignment, and quiz contributes equally toward the final grade.

Extra credit A 2-point extra credit assignment is available on the course website. It provides practice in algebra and is available only during the first half of the semester. The due date is listed in the schedule below.

Quizzes are administered via Canvas and are open book. Most questions will be multiple choice, but you may need to do calculations to choose the right answer.

Weekly computer lab In this lab, students do projects using the Python programming language. The lab assignments are short enough to complete during the two-hour lab. The lab syllabus is available on the class web site. The projects themselves are described in JEPy and in the *Lab Manual for Biol 5221*, which is also available on the website.

Homework There are also paper-and-pencil homework assignments, which are due at roughly weekly intervals as indicated in the schedule below. The homework assignments are available on the class website. Answers to even-numbered problems are in the back of the book of assignments. Only odd-numbered problems will be graded.

Write your homework assignments on paper, with your name and the title of the assignment at the top. Number each page. If there are 3 pages, the numbers should look like “1/3,” “2/3,” and “3/3,” so that we can tell that we have all the pages. Finally, scan the assignment or take a cell-phone photograph, and upload to Canvas. There is a Canvas cell-phone app, will allow you to photograph several pages, one at a time, and upload them all together.

Late assignments If assignments are late, but not more than one week late, we’ll deduct 10% of the grade. We intended not to accept assignments more than one week late, but that policy was not reflected in the syllabus. To keep the promise made in the original syllabus, we’re modifying the policy as follows. For assignments due on or before 9 March 2024, we will accept submissions until the last week of class, with a 50% penalty if they are more than one week late. For assignments due

after 9 March, we will not accept assignments more than one week late. No late assignments will be accepted during the last week of the course. If the assignment is delayed because of a medical problem or other crisis, there is no penalty.

Required readings are listed numerically in the outline below. The full reference corresponding to each number is in the list of references at the end. The main text,

Gillespie, John. 2004. *Population Genetics, a Concise Guide*, 2nd edition

is available at the campus bookstore (also in paper and electronic versions on the web). All other readings are on the class website. In addition, we will occasionally assign other published papers and notes of our own. When we do, they will be available either on paper or on the course web site.

There are also two excellent texts, which are freely available online:

1. Pritchard, Jonathan. 2024 *An Owner's Guide to the Human Genome: An Introduction to Human Population Genetics, Variation and Disease*.
2. Coop, Graham. 2024. *Population and Quantitative Genetics*.

These are not required, but you may find them helpful. We suggest portions of them as optional readings on the course website.

Contact Feel free to contact us either through Canvas or by email. *Hancock* hancock@mpipz.mpg.de; *Rogers*: rogers@anthro.utah.edu; *Seger*: seger@biology.utah.edu.

Learning outcomes

Evolution Students will be able to use the principles of evolutionary theory to interpret patterns of variation within species, and of change over time.

Scientific reasoning Students will be able to apply the method of critical scientific reasoning to identify knowledge gaps, formulate hypotheses, and test them against experimental and observational data to advance an understanding of the natural world.

Quantitative reasoning Students will be able to use mathematical and computational methods and tools to describe living systems and be able to apply quantitative approaches, such as statistics, quantitative analysis of dynamic systems, or mathematical modeling.

Americans with Disabilities Act The University of Utah seeks to provide equal access to its programs, services, and activities for people with disabilities. If you will need accommodations in this class, reasonable prior notice should be given to the Center for Disability and Access, 162 Olpin Union Building, (801) 581-5020.

Attendance Given the nature and goals of this course, attendance is required and adjustments cannot be granted to allow non-attendance. However, if you need to seek an ADA accommodation to request an exception to this attendance policy due to a disability, please contact the Center for Disability and Access (CDA). CDA will work with us to determine what, if any, ADA accommodations are reasonable and appropriate

University Safety Statement The University of Utah values the safety of all campus community members. To report suspicious activity or to request a courtesy escort, call campus police at

801-585-COPS (801-585-2677). You will receive important emergency alerts and safety messages regarding campus safety via text message. For more information regarding safety and to view available training resources, including helpful videos, visit <https://safeu.utah.edu>.

Sexual Misconduct Title IX makes clear that violence and harassment based on sex and gender (including sexual orientation and gender identity/expression) is a civil rights offense subject to the same kinds of accountability and support applied to offenses against other protected categories such as race, national origin, color, religion, age, status as a person with a disability, veteran's status or genetic information. If you or someone you know has been harassed or assaulted, you are encouraged to report it to the Title IX Coordinator in the Office of Equal Opportunity and Affirmative Action, 383 South University Street, 801-581-8365, or the Dean of Students, 270 Union Building, 801-581-7066. For support and confidential consultation contact the Center for Student Wellness, 426 SSB, 801-581-7776. To report to the police, contact the Department of Public Safety, 801-585-2677 (COPS).

Academic Misconduct It is expected that students adhere to University of Utah policies regarding academic honesty, including but not limited to refraining from cheating, plagiarizing, misrepresenting one's work, and/or inappropriately collaborating. This includes the use of generative artificial intelligence (AI) tools without citation, documentation, or authorization. Students are expected to adhere to the prescribed professional and ethical standards of the profession/discipline for which they are preparing. Any student who engages in academic dishonesty or who violates the professional and ethical standards for their profession/discipline may be subject to academic sanctions as per the University of Utah's Student Code.

Schedule

Date	Lecture	Reading
Jan 09 T	Introduction: Scope of course	S
	Probability 1	R [11, sec. 1-2]
11 H	Probability 3	R [11, sec. 3]
16 T	Describing and partitioning phenotypic variation Homework 1 due.	S
18 H	Genomes and their variation	H [3, sec. 1.0-1.3]
23 T	Random mating Decay of heterozygosity	S [3, sec. 1.4] S [3, sec. 2.0-2.2]
25 H	Mutation versus drift	H [3, sec. 2.3]
30 T	Describing DNA sequence variation	S [12, Ch. 1]
Feb 01 H	Gene genealogies Homework 2 due.	R [12, Ch. 4]
06 T	Connecting gene genealogies to genetics Homework 3 due.	R [12, Ch. 5]; [3, sec. 2.6]
08 H	Site frequency spectrum	H [12, Ch. 6]
13 T	Mismatch distribution and population growth	R [12, Ch. 7]
15 H	The neutral theory of molecular evolution 1	S [3, sec. 2.4]
20 T	The neutral theory of molecular evolution 2 Homework 4 due.	S [3, sec. 2.5]
22 H	Selection Extra credit assignment due.	H [3, sec. 3.0-3.3];[15]
27 T	Mutation versus selection	S [3, sec. 3.4]; [2]

	Genetic load	S [3, sec. 3.5]
	Homework 5 due.	
29 H	Fixation of mutations	S [3, sec. 3.9–3.10]
Mar 05 T	*** NO CLASS	
07 H	*** NO CLASS	
12 T	Neutral evolution at two loci	R [3, sec. 4.0–4.1]
	Homework 6 due.	
14 H	Selection at two loci	R [3, sec. 4.2]
	Why LD helps us find selective sweeps	R [9]
19 T	Inbreeding	R [3, sec. 5.0–5.3]
	Homework 7 due.	
21 H	Population structure	H [3, sec. 5.5];[10]
26 T	Detecting natural selection	H [4, 18]
	Homework 8 due.	
28 H	Genetic draft	S [3, sec. 4.3]
Apr 02 T	Population history from whole genomes	R [5]
	Homework 9 due.	
04 H	Archaic genes in modern humans	R [14]
	Selection on archaic genes	R
09 T	Quantitative genetics: genes, environment, heritability	S [3, sec. 6.0–6.2]
	Homework 10 due.	
11 H	Response to selection	S [13]
16 T	Trait mapping	H [16, 17]
	Homework 11 due.	
18 H	Finding the missing heritability: trait prediction and prospects for personalized medicine	H [6–8]
23 T	Evolution of quantitative traits	H [1]
	Homework 12 due.	

References

- [1] Neda Barghi, Joachim Hermisson, and Christian Schlötterer. “Polygenic adaptation: a unifying framework to understand positive selection”. *Nature Reviews Genetics* 21.12 (2020), pp. 769–781. DOI: 10.1038/s41576-020-0250-z.
- [2] Wenqing Fu et al. “Analysis of 6,515 exomes reveals the recent origin of most human protein-coding variants”. *Nature* 493 (2012), pp. 216–270. DOI: 10.1038/nature11690.
- [3] John H. Gillespie. *Population Genetics: A Concise Guide*. 2nd. Baltimore: Johns Hopkins University Press, 2004.
- [4] Hussein A. Hejase, Noah Dukler, and Adam Siepel. “Summary statistics to gene trees: methods for inferring positive selection”. *Trends in Genetics* 36.4 (2020), pp. 243–258. DOI: 10.1016/j.tig.2019.12.008.
- [5] Heng Li and Richard Durbin. “Inference of human population history from individual whole-genome sequences”. *Nature* 475.7357 (2011), pp. 493–496. DOI: 10.1038/nature10231.

- [6] Teri A. Manolio et al. “Finding the missing heritability of complex diseases”. *Nature* 461.7265 (2009), pp. 747–753. DOI: 10.1038/nature08494.
- [7] Arya Massarat et al. “Human pangenome supports analysis of complex genomic regions”. *Nature* 617 (2023), p. 256. eprint: <https://www.nature.com/articles/d41586-023-01490-3.pdf>.
- [8] National Human Genome Research Institute. *Polygenic Risk Scores*. 2024. URL: <https://www.genome.gov/Health/Genomics-and-Medicine/Polygenic-risk-scores>.
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- [13] Alan R. Rogers. *Response to Natural Selection on a Quantitative Character*. 2022. URL: <https://content.csbs.utah.edu/~rogers/tch/ant5221/unprotected/Rogers-lande.pdf>.
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- [16] Vivian Tam et al. “Benefits and limitations of genome-wide association studies”. *Nature Reviews Genetics* 20.8 (2019), pp. 467–484. DOI: 10.1038/s41576-019-0127-1.
- [17] Emil Uffelmann et al. “Genome-wide association studies”. *Nature Reviews Methods Primers* 1.1 (2021), p. 59. DOI: 10.1038/s43586-021-00056-9.
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