EVANTH 318 - HUMAN EVOLUTIONARY GENETICS

COURSE INFORMATION

Cross-listing: BIOLOGY 318
Curriculum Codes: NS, R
Number of Units: 1
Term & Year: Fall 2021
Day & Time: W&F 3:30-4:45 pm
Location: BioSci 113

Professor's Name: Iman Hamid
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COURSE DESCRIPTION

Millions of years of evolution have shaped the genomes of all living and extinct organisms, including modern humans. This course will take an experiential approach to teaching population genetics that will empower students to explore how evolution can shape genomes in real time. We will relate simulated and empirical results to the expectations of classical theoretical examples and current models of human demography and evolution. Using population genetic simulations, students will be guided through independent and group activities that challenge assumptions and improve intuition for theoretical expectations. Students will test how genomic features like mutation rate, demographic features like migration rate and population size, and evolutionary features like natural selection affect genetic variation over time, using both simulated data and real-world datasets from modern human populations. This course will also provide students an introduction to what is increasingly becoming an essential skill in the field of population genetics: computer programming. No prior computational experience is required. Students should have successfully completed Bio 201 and 202 or equivalent prior to enrolling in this course.

STUDENT LEARNING OBJECTIVES

At the end of this course, successful students will be able to:

1. Develop individual hypotheses based on population genetic theory about how demographic and selective forces shape genomes.
2. Model, design, and run simulations that test predictions of these hypotheses.
3. Compare results of empirical and simulated data to theoretical expectations of genetic variation.
4. Evaluate and communicate results to peers through small group projects and final presentation. Defend hypothesis or explain why predictions were not supported by data.

REQUIRED STUDENT RESOURCES

Course materials all available on Sakai. There is no required textbook for this course.

EVALUATION PROCEDURES & GRADING CRITERIA

ASSIGNMENTS & GRADE BREAKDOWN

Paper discussions	10% of final grade	**A+**: 98 to 100%	**A**: 93 to <98%	**A-**: 90 to <93%
Lab simulations	10% of final grade	**B+**: 88 to <90%	**B**: 83 to <88%	**B-**: 80 to <83%
Group assignments	54% of final grade	**C+**: 78 to <80%	**C**: 73 to <78%	**C-**: 70 to <73%
Final presentation	26% of final grade	**D+**: 68 to <70%	**D**: 63 to <68%	**D-**: 60 to <63%
	100%	**F**: <60%		

GRADING CRITERIA

- **Paper Discussions** (10% of final grade, 10 pts each) – We will have eight (8) paper discussions in which we will discuss research or review articles relevant to human evolutionary genetics. For these assignments, you are expected to have read the paper deeply and come prepared to discuss in class. While reading the articles, try to think of the following guided questions:
  - *What is the author's hypothesis?*
  - *What kind of data did they collect and why?*
  - *Do their results support their hypothesis?*
  - *What are the conclusions?*
  - *How does this article relate to what we have discussed in class?*
  - *What new questions does this article spark for you?*

  In class, I will guide the discussion, but you will be evaluated on thoughtful participation. Discussion questions and points **SHOULD NOT** be clarifying questions. For example, you should try to relate the articles to what we have learned in class, evaluate how well the results support the conclusions, consider whether article does or does not align with your previous understanding of human evolution, or propose how this article might inform a future study. You will either earn full (10 points) or no credit (0 points) for each discussion. This portion of your final grade will be averaged over the eight (8) paper discussions.

- **Lab Simulations** (10% of final grade, 10 pts each) – We will have eight (8) lab SLiM simulation assignments, each graded out of ten (10) points. These are designed to be completed during the allotted lab time, but can be completed as homework and are due before the start of the following class period (via Sakai). You are permitted and expected to work on these in class, and you are encouraged to ask for help from myself or any of your peers. However, you are expected to turn in your own individual work, and plagiarism will be penalized with no credit. To avoid this, you should confirm you fully understand the solution before writing up your own assignment independently.

  These will be evaluated based on completion & functionality (i.e. *Does your simulation do what it is supposed to?*). A complete simulation that performs the assigned task will earn full credit (10 points); a complete simulation that does not perform the assigned task because of a bug/mistake will earn partial credit (5 points); otherwise, a complete simulation that does not perform the assigned task will earn no credit (0 points); an incomplete simulation will earn no credit (0 points). This portion of your final grade will be averaged over the eight (8) lab simulation assignments.

- **Group Assignments** (54% of final grade) – This course will emphasize group collaboration. Groups will be determined in the first lab, and you will stay in the same group over the course of the semester. There are a number of assignments you will be expected to complete with your groups. Percentages represent percentage of final grade unless stated otherwise.
  - **Group Contract & Contract Fulfillment** (14%, 10 pts total) – Each group will draft a contract on group expectations for roles and effort with relation to group assignments and final project/presentation (30% of assignment grade, completion). You will evaluate yourself and your peers on fulfillment of this contract periodically throughout the semester (ungraded evaluations). Your fulfillment of this contract as evaluated by yourself and your peers **at the end of the course** is worth 70% of this assignment grade.

  - **Project List of Papers** (5%, 5 pts total) – Each group will submit a list of at least five (5) research or review articles relevant to their project hypothesis, to be used in their proposal introduction. One (1) point will be awarded for each relevant paper listed, up to a total of five (5) points. Irrelevant articles earn no points.

- **Group Simulations** (10%, 10 pts total) – In your groups, you will write SLiM simulations that test a hypothesis from a list of suggested papers. You will then write a short summary evaluating the results of your simulation and comparing to theoretical or empirical expectations. This portion of your final grade will be averaged over the three (3) group simulation assignments. See Appendix A for rubric & grading criteria.

- **Group Paper Discussion** (5%, 10 pts total) – Each group will lead one paper discussion on an article of their choice. Groups should guide discussions following the model of those from earlier in the semester. See Appendix B for rubric & grading criteria.

  In completing this assignment, groups will also earn free full credit for one (1) paper discussion assignment. All other students in class should treat this as a regular paper discussion assignment.

- **Project Proposal** (20%, 10 pts total) – Groups will submit a written proposal for a final project that tests an independent hypothesis. See Appendix C for rubric & grading criteria.

- **Project Simulation** (10%, 10 pts total) – Groups will submit a SLiM simulation based on their project proposal. See Appendix D for rubric & grading criteria.

- **Final Presentation** (26%, 10 pts total) – Groups will present the results of their project in a 30 min presentation. Groups should put their hypothesis in context of both topics discussed in the course as well as previously published work. Groups should either defend their hypothesis using the results of their simulations & available empirical data, or provide reasonable explanations as to why the results do not support their hypothesis. See Appendix E for rubric & grading criteria.

ABSENCE & LATE POLICIES

Please note that you are responsible for all assignments due and material discussed in classes you cannot attend. Further, a decent proportion of lab time is allotted for groups to work on assignments and final projects. Students who miss multiple classes will see their group evaluation grades suffer as a result.

Assignments are all due **by the start of class via Sakai** whether or not you are present. In the event that you miss class, you should submit a Short Term Incapacitation Form (https://class-absences.trinity.duke.edu/if) or a Notice of Varsity Athletics Participation (https://class-absences.trinity.duke.edu/novap) before the start of class. For missed paper discussions, you may submit questions ahead of class time in lieu of your verbal participation. Late paper discussion questions and lab simulations will not be accepted. For all other assignments, groups will be deducted 20% of the assignment grade per 24 hour window past the deadline.

DIVERSITY AND INCLUSION

As a group, we will strive to create a welcoming and safe environment for students and instructors. In group discussions, we recognize that it is okay for others to have differing opinions from our own, and we can use evidence-based rationale to challenge opposing ideas. We will not tolerate disrespectful or discriminatory behavior in the classroom. Failure to uphold these values will result in disciplinary action.

ABILITY ACCOMMODATION

Students who may need accommodations should contact the Student Disabilities Access Office (see https://access.duke.edu/requests). The earlier accommodations are requested, the quicker they can be implemented to ensure your ability to succeed in this course.

ACADEMIC INTEGRITY

All students are expected to uphold the Duke Community Standard (https://studentaffairs.duke.edu/conduct/about-us/duke-communitystandard):

- I will not lie, cheat, or steal in my academic endeavors.
- I will conduct myself honorably in all my endeavors; and
- I will act if the Standard is compromised.

Additionally, this course emphasizes collaborative group learning and projects. Each student is expected to contribute **equal efforts in group assignments and discussions**. At the start of the course, students will draft a mutual contract for expectations within their group. Effort will be informally evaluated through periodic self- and peer-evaluations, and fulfillment of the group contract will be formally evaluated at the end of the course.

ADDITIONAL RESOURCES

Part of being a successful student is taking care of your personal needs and knowing when to ask for help. We are here to help you succeed. Below are some excellent Duke resources to assist with study skills, mental and physical health and wellness, and community building:

- Academic Resource Center: https://arc.duke.edu/
- TWP Writing Studio: https://twp.duke.edu/twp-writing-studio/
- Student Wellness: https://studentaffairs.duke.edu/wellness
- Counseling and Psychological Services: https://studentaffairs.duke.edu/caps
- Women's Center: https://studentaffairs.duke.edu/wc
- DukeReach: https://studentaffairs.duke.edu/dukereach1
- Blue Devils Care: https://bluedevilscare.duke.edu/
- DuWell: https://studentaffairs.duke.edu/duwell/
- International House: https://studentaffairs.duke.edu/ihouse

COURSE SCHEDULE

We meet bi-weekly; that is, two 75 minute classes per week. Typically, we will spend one class on the lecture topic and the next class doing interactive labs. Later in the semester, we will shift to more frequent paper discussions as student groups hone in on their independent project hypotheses and methods. Lab simulations are designed to be completed during class time, but will be due at the start of next class. *Graded assignments*, ungraded assignments, and **group assignments** are due before class (via Sakai) on the date listed.

Date		Topic	Format	Assignments Due
25-Aug	**W**	Overview: models of populations & evolutionary mechanisms	Lecture	
27-Aug	**F**	Intro to SLiM. How do we simulate populations, genomes, mutations, etc?	Lab	Install SLiM
1-Sep	**W**	How do we define genetic variation? genotypes, alleles, haplotypes	Lecture	*Lab Simulation 1: Population model*
3-Sep	**F**	Explore genotype data, in SLiM & 1000G Group project overview	Lab	
8-Sep	**W**	Measuring genetic variation	Lecture & Lab combined	*Lab Simulation 2: Genomes & mutations*
10-Sep	**F**	Recombination, LD	Lecture & Lab combined	*Lab Simulation 3: SFS* ***Group Contract***
15-Sep	**W**	Population structure & geography	Lecture	*Lab Simulation 4: Recombination Rate & Neutral Variation*
17-Sep	**F**	Paper Discussion: "Genes mirror geography within Europe" – Novembre et al. 2008 *Nature*	Lab	*Lab Simulation 5: Calculate Fst* *Paper Discussion 1*
22-Sep	**W**	Population growth history & measures of genetic variation	Lecture	***Project List of Papers***
24-Sep	**F**	Models of human evolution, compare/contrast results	Lab	
29-Sep	**W**	Selection	Lecture	**Project Hypothesis**
1-Oct	**F**	Modeling selection in SLiM; calculating common tests/statistics	Lab	
6-Oct	**W**	Paper Discussion: "Population Genomics of Human Adaptation" – Lachance & Tishkoff 2013 *Annu. Rev. Ecol. Evol. Syst*	Lab	*Lab Simulation 6: Selection statistics* *Paper Discussion 2*

8-Oct	**F**	Migration, admixture	Lecture	***Group Simulation 1: OOA simulations & empirical data***
13-Oct	**W**	Admixture & selection	Lecture	*Lab Simulation 7: Gene flow*
15-Oct	**F**	Combining selection & demography in SLiM models	Lab	**Group Effort Evaluation 1**
20-Oct	**W**	Genome-wide scans for selection	Lecture	*Lab Simulation 8: Selection & Demography* **Project proposal draft**
22-Oct	**F**	Disease genetics	Lecture	***Group Simulation 2: Adaptive introgression & empirical data***
27-Oct	**W**	GWAS examples	Lab	**Group Effort Evaluation 2**
29-Oct	**F**	Genetics & society: human genomes, representation, and equity. Paper Discussion: "Genomics is failing on diversity" – Popejoy & Fullerton 2016 *Nature News & Comment*	Lecture & Lab combined	*Paper Discussion 3*
3-Nov	**W**	Genetics & society: genealogical vs genetic ancestry. Direct to consumer ancestry testing (e.g. 23&me & AncestryDNA) Paper discussion: "What is Ancestry?" Mathieson & Scally 2020 *PLoS Genetics*	Lecture & Lab combined	***Group Simulation 3: Changing environment*** *Paper Discussion 4*
5-Nov	**F**	Genetics & society: history of eugenics in our field	Lab	***Project proposal***
10-Nov	**W**	Paper Discussion: ***Group 1***	Lab	**Group Effort Evaluation 3** *Paper Discussion 5*
12-Nov	**F**	Paper Discussion: ***Group 2***	Lab	*Paper Discussion 6*
17-Nov	**W**	Paper Discussion: ***Group 3***	Lab	*Paper Discussion 7*
19-Nov	**F**	Paper Discussion: ***Group 4***	Lab	*Paper Discussion 8* ***Project simulation***
24-26-Nov	**W & F**	Thanksgiving break	-	-
W 12/1	**W**	***Group 1 & 2*** presentations	-	***Project Presentations***
F 12/3	**F**	***Group 3 & 4*** presentations	-	***Final Group Effort Evaluation*** ***Project Presentations***

APPENDIX A: GROUP SIMULATION RUBRIC

Grades will be determined by the instructor according to these guidelines (max 10 pts, weighted total):

- Is the assignment completed? (5% of assignment grade)
- Did the group accurately identify the original article's hypothesis? (5% of assignment grade)
- Is the simulation designed to test the hypothesis of interest? (35% of assignment grade)
- Does the simulation work as intended? (20% of assignment grade)
- Does the written summary accurately evaluate results? (35% of assignment grade)

	Full Credit (10 pts)	**Partial Credit** (5 pts)	**No Credit** (0 pts)
Completion *5% assignment grade*	Assignment is completed. Group submitted a full simulation and written summary, including a description of the hypothesis tested and evaluation of simulated results.	Assignment is not fully completed. Group submitted either a partial simulation OR a partial written summary.	Assignment is not completed. Group did not submit simulation and/or written summary OR group submitted both the written summary and simulation partially completed.
Hypothesis *5% assignment grade*	Group accurately identified the original article's hypothesis.		Group did not accurately identify the original article's hypothesis.
Simulation Design *35% assignment grade*	Simulation is designed to test the hypothesis of interest. The model and data are appropriate for the question.	Simulation does not fully test the hypothesis of interest. Either the model OR the data are not appropriate for the question.	Simulation does not test the hypothesis of interest. Simulation is either off-topic, incomplete, or the model AND the data are not appropriate for the question.
Simulation Functionality *20% assignment grade*	The simulation works as intended. There are no errors, and the output is as expected.	The simulation does not fully work as intended. There may be up to two minor bugs that stop the simulation before completion OR the simulation completes, but the results are not as expected due to an error.	The simulation does not work as intended. There are more than two bugs that stop the simulation before completion or the written simulation is incomplete.
Results Evaluation *35% assignment grade*	Written summary accurately evaluates results of simulation. Simulation results are explained in context of course topics and are visualized appropriately. If results do not support the hypothesis, a reasonable explanation is provided.	Written summary inaccurately evaluates results of simulation OR results are not interpreted in the context of course topics OR simulation results may not be explained and/or are not visualized appropriately. If results do not support the hypothesis, an explanation is provided, though it may be unreasonable.	Written summary inaccurately evaluates results of simulation AND results are not interpreted in the context of course topics OR simulation results are not explained or are not visualized appropriately. If results do not support the hypothesis, an explanation is either not provided or is unreasonable.

APPENDIX B: GROUP PAPER DISCUSSION

Grades will be determined by the instructor according to these guidelines (max 10 pts, weighted total):

- Did the group accurately convey the article's hypothesis? (25% of assignment grade)
- Did the group accurately convey the results of the article? (25% of assignment grade)
- Did the group relate the article to topics learned in class? (25% of assignment grade)
- Did the group ask thought-provoking questions about the article's results and conclusions? (25% of assignment grade)

	Full Credit (10 pts)	**Partial Credit** (5 pts)	**No Credit** (0 pts)
Hypothesis *25% of assignment grade*	Group accurately and clearly conveys the article's hypothesis. There are no or one clarifying question.	Group did not convey the article's hypothesis accurately OR clearly. There may be more than one clarifying question about the hypothesis.	Group did not convey the article's hypothesis at all OR did so inaccurately. There are many clarifying questions and/or the instructor had to provide clarification.
Results *25% of assignment grade*	Group accurately and clearly conveyed most of the results of the article. There are few clarifying questions.	Group did not convey all the results accurately or clearly. There are many clarifying questions.	The group conveys the most of the article's results inaccurately OR instructor had to provide clarification more than once.
Context *25% of assignment grade*	Group related the article to topics learned in class.	Group related the article to topics learned in class, but inaccurately or inappropriately.	Group does not relate the article to topics learned in class.
Discussion *25% of assignment grade*	Group asks many thought-provoking questions about the articles results and conclusions.	Group asks fewer than three thought-provoking questions about the results and conclusions	Group asks no thought-provoking questions about the results and conclusions.

APPENDIX C: PROJECT PROPOSAL RUBRIC

Grades will be determined by the instructor according to these guidelines (max 10 pts, weighted total):

- Has the group developed a hypothesis about how demographic or selective forces shape genomic variation? (20% of assignment grade)
- Does the hypothesis relate to and build on topics discussed in course? (10% of assignment grade)
- Is the hypothesis testable? (10% of assignment grade)
- Do the proposal introduction & cited papers justify the hypothesis? (15% of assignment grade)
- Will the proposed method test the hypothesis? (25% of assignment grade)
- Is the proposed method feasible? (10% of assignment grade)
- Is the proposed data appropriate? (10% of assignment grade)

See next page for rubric.

APPENDIX C: PROJECT PROPOSAL RUBRIC – CONT'D

		Full Credit (10 pts)	**Partial Credit** (5 pts)	**No Credit** (0 pts)
Hypothesis *40% of assignment grade*	**Topic** *20% of assignment grade*	Group has developed a clear hypothesis on how demographic and/or selective forces shape genomic variation. There is no ambiguity about group's predictions.	Group has developed a hypothesis on how demographic and/or selective forces shape genomic variation, but it is not clear. There may be ambiguity about the group's hypothesis or predictions.	Group has not developed a hypothesis at all OR it is not relevant to the effects of demography and/or selection.
	Course context *10% of assignment grade*	Group's hypothesis relates to and builds on topics discussed in the course.	Group's hypothesis relates to topics discussed in class, but does not build beyond that to constitute independent development.	Group has not developed a hypothesis at all OR group's hypothesis is unrelated to course topics.
	Testability *10% of assignment grade*	Group's hypothesis is testable given what we have learned in class.	Group's hypothesis is testable, but not using the tools we have covered in class.	Group's hypothesis is not testable.
Introduction *15% of assignment grade*		Introduction and cited papers clearly justify the hypothesis. At least 5 papers are cited. Reasoning for proposed hypothesis is thoroughly explained.	Introduction and cited papers do not clearly justify the hypothesis. Fewer than 5 papers are cited OR reasoning for proposed hypothesis is not thoroughly explained.	Introduction does not clearly justify the hypothesis. Less than 2 papers are cited, and there is no or very little reasoning for proposed hypothesis.
Methods *35% of assignment grade*	**Applicability** *25% of assignment grade*	The proposed method will test the group's hypothesis. The method and model are appropriate for the question.	The proposed method will partially test the group's hypothesis, but there are important aspects of the hypothesis not addressed by the method. Model may be inappropriate for the question.	The proposed method will not test the group's hypothesis at all. The model is inappropriate for the question.
	Feasibility *10% of assignment grade*	The proposed method is feasible and can be completed in the given time frame using the tools we have learned in class. The group can proceed as proposed.	The proposed method is feasible, but may need some tweaking to be completed in the given time frame or using the tools we have learned in class.	The proposed method is not feasible given the time frame or the tools we have learned in class. The group will need to completely re-evaluate their proposed methods.
	Data *10% of assignment grade*	The proposed data is appropriate for the question, and there are no or few concerns about whether results can be interpreted in context of the hypothesis.	The proposed data is appropriate for the question, but there are concerns whether the results may lead to alternate interpretations.	The proposed data is inappropriate for the question. Results would not provide adequate support for the hypothesis.

APPENDIX D: PROJECT SIMULATION RUBRIC

Grades will be determined by the instructor according to these guidelines (max 10 pts, weighted total):

- Does the simulation work as intended? (30% of assignment grade)
- Does the simulation test the proposed hypothesis? (70% of assignment grade)

	Full Credit (10 pts)	**Partial Credit** (5 pts)	**No Credit** (0 pts)
Simulation Functionality *30% assignment grade*	The simulation works as intended. There are no errors, and the output is as expected.	The simulation does not fully work as intended. There may be one minor bug that stops the simulation before completion OR the simulation completes, but the results are not as expected due to an error.	The simulation does not work as intended. There is more than one bugs that stops the simulation before completion or the written simulation is incomplete.
Simulation Design *70% assignment grade*	Simulation is designed to test the proposed hypothesis. The model and data are appropriate for the question. If group project proposal was returned with concerns about methods, all concerns have been rectified in this simulation design.	Simulation does not fully test the hypothesis of interest. Either the model OR the data are not appropriate for the question. If group project proposal was returned with concerns about methods, some concerns have not been rectified.	Simulation does not test the hypothesis of interest. Simulation is either off-topic, incomplete, or the model AND the data are not appropriate for the question. If group project proposal was returned with concerns about methods, many concerns have not been rectified.

APPENDIX E: FINAL PRESENTATION RUBRIC

Grades will be determined by the instructor according to these guidelines (max 10 pts, weighted total):

- Did the group provide convincing & accurate background to justify their hypothesis? (10% of assignment grade)
- Did the group accurately evaluate their results? (30% of assignment grade)
- Did the group successfully explain whether their results support their hypothesis and why/why not? (40% of assignment grade)
- Did peers feel the group effectively communicated their results & conclusions to the class? (20% of assignment grade)

	Full Credit (10 pts)	**Partial Credit** (5 pts)	**No Credit** (0 pts)
Background *10% of assignment grade*	Group provided convincing and accurate background to justify their hypothesis. Groups put their hypothesis in context of both topics discussed in course as well as previously published work.	Group provided accurate background to justify their hypothesis but it may have been unconvincing. Background is put in context of topics discussed in course but may not have included previously published work	Group provided inaccurate or inadequate background; hypothesis is not justified. Relevant course topics and previously published work are discussed briefly or not at all.
Results *30% of assignment grade*	The group accurately evaluated and interpreted their results. The group explains the results well, and demonstrates a clear understanding of relevant course topics.	The group evaluated their results, but there are some inaccuracies or inappropriate interpretations. Some results were not explained well.	The group did not evaluate their results. The group may have listed results without interpretation or explanation. Results may have been explained poorly, demonstrating misunderstanding of course topics.
Conclusion *40% of assignment grade*	Group successfully defended their hypothesis using results of their simulated and/or available empirical data OR group has provided reasonable explanations as to why the results do not support their hypothesis, synthesizing the knowledge they have learned in the course.	Group defends their hypothesis using the results of their simulation OR provides an explanation as to why the results do not support their hypothesis, but there are inaccuracies in their conclusions. Group may have difficulty using course topics to explain how/why the results do or do not support their hypothesis.	Group does not relate the results back to their hypothesis, or completely misinterprets the results. Group does not use knowledge gained from course topics to explain how/why the results do or do not support their hypothesis.
Peer Evaluation *20% of assignment grade*	Peers in the audience felt the group effectively communicated their results and conclusions to the class. There were few clarifying questions or concerns about the background, method, hypothesis, and interpretations of results and final conclusions.	Peers in the audience felt the group adequately presented their results and conclusions to the class, but there were many clarifying questions and a few concerns about the validity of results and final conclusions.	Peers in the audience had difficulty following the presentation, there were many concerns about the validity or relevance of topics discussed.