Evolutionary Genetics Parul Johri, Ph.D. BIOL 454-002

Office hours: On request.

## Text (optional):

Elements of Evolutionary Genetics by Brian Charlesworth & Deborah Charlesworth Population genetics: a concise guide by John H. Gillespie

**Course description**: The roles of mutation, genetic drift, population history, and natural selection in the evolution of the genotype. Basic principles are applied to biological studies. Three lecture hours a week.

**Requisites:** Prerequisites, BIOL 201 and BIOL 202; or BIOL 103 and BIOL 104; or permission of the instructor for students lacking the prerequisites.

Course Goals: The aim of the course is to provide an overview of the evolutionary processes that shape genomic variation. The course will have a heavy quantitative emphasis and you will be challenged to understand derivations, experiments, conclusions, and primary literature.

Set up in canvas

# **CLASS HOURS AND VENUE**

Fall 2023

M: Monday, W: Wednesday, F: Friday. 10:10-11:00 am Genome Sciences Building, Room 1374

# **SYLLABUS**

# **MODULE 1. Basic evolutionary processes**

# Week 1: Genetic variation [Aug 21 - 25]

M: Discussion of the syllabus + Introduction to population genetics (lecture)

W: Genetic variation + Basic probability + Hardy Weinberg Equilibrium (lecture)

F: Problem Set #1

# Week 2: Mutation and Genetic drift I [Aug 28 – Sep 1]

M: Genetic Drift (Lecture)

W. Mutation (Lecture)

# F: Problem Set #2

# Week 3: Mutation and Genetic drift II

M: No class

W: Paper presentation #1

<u>F</u>: Paper presentation #2

# Week 4: Selection [Sep 11 -15]

M: Selection at single sites (lecture)

W: Problem Set #3

<u>F</u>: Paper presentation #3

# Week 5: Recombination [Sep 18-22]

<u>M</u>: Linkage disequilibrium (Lecture)

W: Problem Set #4

F: Paper presentation #4

# Week 6: Review + Midterm [Sep 25-29]

M: No class (Wellness Day)

W: Class open to questions

F: MIDTERM #1

# **MODULE 2. Complex evolutionary processes**

# Week 7: Coalescence Theory [Oct 2-6]

M: Lecture

W: Problem Set #5

<u>F</u>: Paper presentation #5 [Chapter 1, Coalescent Theory by John Wakeley]

# Week 8: Population structure [Oct 9 – 13]

M: Lecture

W: Problem set #6

F: Paper presentation #6

# Week 9: Demographic history [Oct 16- 20]

M: Lecture

W: Paper presentation #7

F: No class (Fall Break)

# **Module 3: EMPIRICAL APPLICATIONS/ ADVANCED CONCEPTS**

# Week 10: Midterm + Heterogeneity in mutation and recombination rates [Oct 23-27]

M: MIDTERM #2 (material from Module 2)

W: Group Reading #1

F: Paper presentation #8

# Week 11: The distribution of fitness effects of new mutations [Oct 30 – Nov 3]

**M**: Estimation of the distribution of fitness effects + MK test (Lecture)

W: Group Reading #2

**F:** Paper presentation #9

# Week 12: Effects of selection on linked sites [Nov 6 - 10]

**M**: Hill-Robertson Effects: Background selection + selective sweeps + AOD + Muller's ratchet (Lecture)

W: Group Reading #3

**F:** Paper presentation #10

# Week 13: Sequencing and genotyping errors [Nov 13 – 17]

**M**: Genotyping errors and effects on summary statistics (Lecture)

W: Group Reading #4

**F:** Paper presentation #11

# Week 14: Heritability [Nov 20]

M: Group Reading #5 – Chapter 3, Section 3.3 from Ch & Ch (Pages 114-129)

W: No Class

F: No Class

# Week 15: Genome Evolution [Nov 27 – Dec 1]

<u>M</u>: Other mutation types - transposable elements, gene duplications (Lecture)

W: Group Reading #6

F: Paper presentation #12

# Week 16: Last lecture and review [Dec 4, 6]

M: Open questions or debates in population genetics (Lecture)

W: Class open to questions

# **FINAL EXAM**

# **Grading Scale:**

$$A = 92.9 - 100$$
  $C + = 77.9 - 79.8$   $F = 0 - 59.8$ 

# **Grading**

Problem Set/Group Readings: 10% (participation) + 10% (speaking or presenting)

Paper questionnaire: 20% Paper presentation: 20% Midterm exam #1: 10% Midterm exam #2: 10%

Final Exam: 20%

#### **GRADES**

## Participation in Problem Set/Group Reading:

On Wednesdays, we will be solving problem sets or discussing a review paper in small groups. After the discussion within groups, each group will present what they discussed. You must make sure that you present at least once.

The problem sets will be released during class.

For group reading, you must read the assigned reading before coming to class.

# **Paper Presentations**:

Each week (usually on Fridays) a group of 2-3 students will present and lead the discussion of a weekly paper (posted on canvas). Each student must sign up to present one paper. Sign up for the papers at:

 $\frac{https://docs.google.com/spreadsheets/d/1TWF\_wUMoKPRw6EyWnlNyvGgyevJZt2dyfbSrKDe\_p7OU/edit?usp=sharing}{}$ 

The presenters are also responsible for leading the discussion of the paper.

#### Paper questionnaire

For each paper presentation (usually held on Fridays), each student must submit a questionnaire on Canvas. The reading questionnaire is due on Friday before class.

#### Midterm exams

There will be two midterm exams.

Midterm 1 will include material only from Module 1.

Midterm 2 will include material only from Module 2.

#### Final exam:

Final Exam will include material from the whole semester.

<u>Grade Complaints</u>: Re-grade requests are allowed. Such requests must be made in within one week of receiving a grade and they must be made in written form. I will consider it and make the final decision.

#### **TECHNOLOGY USE**

- Use of computers is allowed but only for taking notes and solving problem sets.
   The use of facebook, twitter, youtube or any other social media during class is prohibited.
- Recording and distributions of lectures is prohibited.
- Use of cell phones and similar devices is prohibited.

## Syllabus Changes

The instructor reserves the right to make changes to the syllabus including project due dates and test dates. These changes will be announced as early as possible.

## **Attendance Policy**

**University Policy:** As stated in the University's <u>Class Attendance Policy</u>, no right or privilege exists that permits a student to be absent from any class meetings, except for these University Approved Absences:

- Authorized University activities: <u>University Approved Absence Office (UAAO)</u>
   <u>website</u> provides information and <u>FAQs for students</u> and <u>FAQs for faculty</u> related to University Approved Absences
- Disability/religious observance/pregnancy, as required by law and approved by <u>Accessibility Resources and Service</u> and/or the <u>Equal Opportunity and</u> <u>Compliance Office</u> (EOC)
- 3. Significant health condition and/or personal/family emergency as approved by the Office of the Dean of Students, Gender Violence Service Coordinators, and/or the Equal Opportunity and Compliance Office (EOC).

Instructors may work with students to meet attendance needs that do not fall within University approved absences. For situations when an absence is not University approved (e.g., a job interview, illness/ flu or club activity), instructors are encouraged to work directly with students to determine the best approach to missed classes and make-up assessment and assignments.

#### **Honor Code**

All students are expected to follow the guidelines of the UNC Honor Code. In particular, students are expected to refrain from "lying, cheating, or stealing" in the academic context. If you are unsure about which actions violate the Honor Code, please see me or consult studentconduct.unc.edu.

#### Acceptable Use Policy

By attending the University of North Carolina at Chapel Hill, you agree to abide by the University of North Carolina at Chapel Hill policies related to the acceptable use of IT systems and services. The Acceptable Use Policy (AUP) sets the expectation that you

will use the University's technology resources responsibly, consistent with the University's mission. In the context of a class, it's quite likely you will participate in online activities that could include personal information about you or your peers, and the AUP addresses your obligations to protect the privacy of class participants. In addition, the AUP addresses matters of others' intellectual property, including copyright. These are only a couple of typical examples, so you should consult the full <a href="Information Technology Acceptable Use Policy">Information Technology Acceptable Use Policy</a>, which covers topics related to using digital resources, such as privacy, confidentiality, and intellectual property. Additionally, consult the <a href="Safe Computing at UNC">Safe Computing at UNC</a> website for information about data security policies, updates, and tips on keeping your identity, information, and devices safe.

# Accessibility Resources and Service

Accessibility Resources and Service (ARS – ars@unc.edu) receives requests for accommodations, and through the Student and Applicant Accommodations Policy determines eligibility and identifies reasonable accommodations for students with disabilities and/or chronic medical conditions to mitigate or remove the barriers experienced in accessing University courses, programs and activities.

ARS also offers its Testing Center resources to students and instructors to facilitate the implementation of testing accommodations.

Faculty and instructors with any concerns or questions about accommodations and/or their implementation, are invited to reach out to ARS to discuss.

# **Counseling and Psychological Services**

UNC-Chapel Hill is strongly committed to addressing the mental health needs of a diverse student body. The <a href="Heels Care Network">Heels Care Network</a> website is a place to access the many mental health resources at Carolina. CAPS is the primary mental health provider for students, offering timely access to consultation and connection to clinically appropriate services. Go to their website <a href="https://caps.unc.edu/">https://caps.unc.edu/</a> or visit their facilities on the third floor of the Campus Health building for an initial evaluation to learn more. Students can also call CAPS 24/7 at 919-966-3658 for immediate assistance.

#### Title IX and Related Resources

Any student who is impacted by discrimination, harassment, interpersonal (relationship) violence, sexual violence, sexual exploitation, or stalking is encouraged to seek resources on campus or in the community. Reports can be made online to the EOC at <a href="https://eoc.unc.edu/report-an-incident/">https://eoc.unc.edu/report-an-incident/</a> or by contacting the University's Title IX Coordinator (Elizabeth Hall, <a href="mailto:titleixcoordinator@unc.edu">titleixcoordinator@unc.edu</a>) or the Report and Response Coordinators in the Equal Opportunity and Compliance Office (<a href="mailto:reportandresponse@unc.edu">reportandresponse@unc.edu</a>). Confidential resources include Counseling and Psychological Services and the Gender Violence Services Coordinators (<a href="mailto:gysc@unc.edu">gysc@unc.edu</a>). Additional resources are available at <a href="mailto:safe.unc.edu">safe.unc.edu</a>.

#### Policy on Non-Discrimination

The University is committed to providing an inclusive and welcoming environment for all members of our community and to ensuring that educational and employment decisions are based on individuals' abilities and qualifications. Consistent with this principle and applicable laws, the University's <u>Policy Statement on Non-Discrimination</u> offers access

to its educational programs and activities as well as employment terms and conditions without respect to race, color, gender, national origin, age, religion, genetic information, disability, veteran's status, sexual orientation, gender identity or gender expression. Such a policy ensures that only relevant factors are considered, and that equitable and consistent standards of conduct and performance are applied.

If you are experiencing harassment or discrimination, you can seek assistance and file a report through the Report and Response Coordinators (email <a href="mailto:reportandresponse@unc.edu">reportandresponse@unc.edu</a> or see additional contact info at <a href="mailto:safe.unc.edu">safe.unc.edu</a>) or the Equal Opportunity and Compliance Office at <a href="https://eoc.unc.edu/report-an-incident/">https://eoc.unc.edu/report-an-incident/</a>.

# **Undergraduate Testing Center**

The College of Arts and Sciences provides a secure, proctored environment in which exams can be taken. The center works with instructors to proctor exams for their undergraduate students who are not registered with ARS and who do not need testing accommodations as provided by ARS. In other words, the Center provides a proctored testing environment for students who are unable to take an exam at the normally scheduled time (with pre-arrangement by your instructor). For more information, visit <a href="http://testingcenter.web.unc.edu/">http://testingcenter.web.unc.edu/</a>.

# **Learning Center**

Want to get the most out of this course or others this semester? Visit UNC's Learning Center at <a href="http://learningcenter.unc.edu">http://learningcenter.unc.edu</a> to make an appointment or register for an event. Their free, popular programs will help you optimize your academic performance. Try academic coaching, peer tutoring, STEM support, ADHD/LD services, workshops and study camps, or review tips and tools available on the website.

# **Writing Center**

For free feedback on any course writing projects, check out UNC's Writing Center. Writing Center coaches can assist with any writing project, including multimedia projects and application essays, at any stage of the writing process. You don't even need a draft to come visit. To schedule a 45-minute appointment, review quick tips, or request written feedback online, visit <a href="http://writingcenter.unc.edu">http://writingcenter.unc.edu</a>.

#### **Syllabus Guidelines for Generative Al**

# Generative AI is extremely useful; however, it has the following limitations:

- How output is arrived at is not clear as the internal processes used to produce a particular output within the generative AI cannot be determined.
- The output is based on existing data (often scraped from online sources) and may reflect biases that should be acknowledged; it may also be inaccurate or entirely fabricated, even if it appears reliable or factual.
- Al evokes a range of intellectual property concerns; sourcing and ownership of information is unclear, and the status of Al output raises numerous questions—e.g., is output equivalent to a published resource? What citational responsibilities are in place for various Al interactions?

The following sections provide the philosophy and specific guidelines for using these tools and features (increasingly, generative AI capabilities will be integrated with

everyday applications). Unless I provide other guidelines for an assignment or exam, you should follow these guidelines.

## **Usage Philosophy**

# Use/disuse of generative AI in your coursework is based on the following principles:

- 1. Al should help you think. Not think for you.
  - Use these tools to give you ideas, perform research (in compliance with point 2 below), and analyze problems. Do not use them to do your work for you, e.g., do not enter an assignment question into ChatGPT and copy & paste the response as your answer.
- 2. Engage with Al Responsibly and Ethically: Engage with Al technologies responsibly, critically evaluating Al-generated outputs and considering potential biases, limitations, and ethical implications in your analysis and discussions. Utilize Al technologies ethically, respecting privacy, confidentiality, and intellectual property rights. Ensure that the data used for Al applications is obtained and shared responsibly and in compliance with relevant regulations.
- 3. You are 100% responsible for your final product.
  You are the user. If the AI makes a mistake, and you use it, it's your mistake. If you don't know whether a statement about any item in the output is true, then your responsibility is to research it. If you cannot verify it as factual, you should delete it. You hold full responsibility for AI-generated content as if you had produced the materials yourself. This means ideas must be attributed, facts are true, and sources must be verified.
- These guidelines are in effect unless I give you specific guidelines for an assignment or exam. It is your responsibility to ensure you are following the correct guidelines.
- 5. Data that are confidential or personal should not be entered into generative

Putting confidential or personal data (e.g., your One Card details) into these tools exposes you and others to the loss of important information. Therefore, do not do so.

The following sections provide the philosophy and specific guidelines for using these tools and features (increasingly, generative AI capabilities will be integrated with everyday applications). **Unless I provide other guidelines for an assignment or exam, you should follow these guidelines.** 

#### **Guideline Specifics**

Not following these guidelines may be a reportable violation to the UNC Honor Court.

Use of generative AI in your coursework is strongly discouraged unless specified otherwise. Note that you are 100% responsible for your final product. You hold full responsibility for AI-generated content as if you had produced the materials yourself.

# **Assignments**

- Readings and Discussions: Do not generate responses to readings using Al.
- Written & Oral Exams: The utilization of AI tools is prohibited and could potentially constitute a reportable violation to the UNC Honor Court.