

PHC 6088: Statistical Analysis of Genetic Data (3 credit hours)

Semester: Spring 2025
Delivery Format: On-Campus

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Office Hours: Mondays 2-3pm and by appointment
Preferred Course Communications: e-mail or Canvas message

Prerequisites

Prior statistical training via PHC6092 and PHC6050c (or equivalent). Permission at the discretion of the instructor may be granted if the student is familiar with linear algebra, maximum likelihood, simple hypothesis testing, and linear regression. There are no prerequisites in genetics; an overview of relevant genetic concepts will be covered in class. R programming experience is helpful, but not required.

Purpose and Outcome

Course Overview

An introduction to statistical procedures for genetic studies. Topics that will be covered include basic population/quantitative genetic concepts (Hardy-Weinberg Equilibrium, linkage disequilibrium, additive/dominant genetic models), QTL mapping, linkage analysis, and genome-wide association studies. This class will emphasize the statistical theory behind methods for analyzing genetic data and its application in useful software tools. The goal of this course is to prepare students for potential research in statistical genetics but is also open to a wider community.

Relation to Program Outcomes

To introduce a variety of statistical methods commonly used in analyzing genetic data, with a focus on linkage analysis, disease mapping, and association studies.

Course Objectives and/or Goals

Upon successfully completing this course, students should be able to:

1. Describe classical genetic concepts such as chromosomal theory of inheritance and meiotic recombination.
2. Discuss basic population and quantitative genetic principles such as Hardy-Weinberg Equilibrium and be able to estimate allele and genotype frequencies.

3. Discuss the difference between linkage and association studies, and family-based versus population-based studies.
4. Describe differences in statistical methods for QTL mapping in experimental crosses (single-marker, EM, regression).
5. Understand how to perform QTL and GWAS analyses via widely-used software platforms (R/plink).

Instructional Methods

Mondays will be one period of lecture. Weekly quizzes due by 12:30pm Monday will assess comprehension of the previous week’s material. Wednesday will be a one period lecture followed by a one period problem-solving session involving problem-sets or project development. Assessments will take place on Wednesdays in place of problem-sets.

Description of Course Content

Topical Outline/Course Schedule

Instructor reserves the right to modify the course schedule with advance notice provided to students.

Week	Date(s)	Topic(s)	Assignments
1	1/13, 1/15	Introduction to genetics	
2	1/22	Introduction to R; genetic data simulations	Assessment 1
3	1/27, 1/29	Quantitative genetics and genetic models	Problem set 1 (in-class)
4	2/3, 2/5	Quantitative trait loci (QTL) mapping	
5	2/10, 2/12	QTL data analysis	Problem set 2 (in-class)
6	2/17, 2/19	Multiple QTL mapping	
7	2/24, 2/26	Relatedness; family-based designs	Problem set 3 (in-class)
8	3/3, 3/5	Population-based designs; GWAS	
9	3/10, 3/12	Meta-analysis for GWAS	Assessment 2
--- Spring Break – No Class ---			
10	3/24, 3/26	GWAS data analysis	Project proposal
11	3/31, 4/2	Genome-wide heritability	
12	4/7, 4/9	Rare-variant analysis	
13	4/14, 4/16	Genomic risk scores (GRS)	
14	4/21, 4/23	Student project presentations	Project presentations

Course Materials and Technology

There is no required text. Instead, lecture notes will be given out over the course of the semester.

Helpful textbooks include:

Mills, Melinda C., Nicola Barban, and Felix C. Tropf. *An introduction to statistical genetic data analysis*. Mit Press, 2020.

Falconer, D. S., and T. F. C. Mackay. *Introduction to quantitative genetics 4th edition*. Pearson, Prentice Hall, 2009.

Lynch, Michael, and Bruce Walsh. *Genetics and analysis of quantitative traits*. Vol. 1. Sunderland, MA: Sinauer, 1998.

Course materials will be available through the Canvas course website at <https://ufl.instructure.com>. It is imperative that students familiarize themselves with Canvas, check Canvas frequently for possible announcements, and make sure that their e-mail account in Canvas is correct and active.

Students will be required to use their own computers in order to complete the assignments, and homework problems will require R programming. R is freely available to download on all operating systems at <https://cran.r-project.org/>. RStudio Desktop is a free, easy-to-use interface to R and highly recommended (<https://posit.co/download/rstudio-desktop/>).

For technical support for this class, please contact the UF Help Desk at:

- Learning-support@ufl.edu
- (352) 392-HELP - select option 2
- <https://lss.at.ufl.edu/help.shtml>

Academic Requirements and Grading

All assignments must be typed (unless otherwise noted) and submitted electronically in Canvas. Your responses must be supported by both written explanations and the code you generate to produce your result.

Weekly quizzes (Online) (5%) – A brief question will be posed at the beginning of each week to ensure students comprehension of the previous week’s material. Two attempts are permitted to complete each quiz and the highest score will be recorded. Quizzes are due by 12:30pm on Monday via Canvas.

Problem sets/Participation (15% / 5%) – There are three problem sets assigned throughout the course. These will largely be completed *in-class*. Students will be able to work in groups, and assignments must be submitted by the announced deadline to receive full-credit. A typical assignment will include a variety of problems. Students may be asked to: calculate or estimate various statistics, simulate data from a particular statistical model and vary initial parameter settings or compare models, derive new estimates of statistics used in genetic studies and compare to ones derived in class, critically examine relevant literature. For problems involving calculations, all work

(or code) must be shown to receive full credit. For problems involving comparisons of models or examinations of the literature, questions are written to elicit thoughtful responses (e.g. questions starting with Why?). *Discussion is allowed, but plagiarism is prohibited. Students must submit their own assignments, written in their own words. Copying of code or explanations is prohibited and will warrant a score of zero.* Solutions will be reviewed in class.

Students exhibiting superior participation will be prepared for class, actively participate in class discussions, and ask questions. The success of an active learning environment depends upon the engagement of students.

Assessments (35%) – Two in-class assessments will be given. The first assessment (10%) covers the genetic concepts to ensure students understand the basics of genetics as presented in course notes. The second assessment (25%) will cover subsequent material covered in class.

Course Project (40%) – The goal of the project is to complete a genetic data analysis from start to finish. Students should download publicly available genetic data and re-analyze the data differently than the original authors (or subsequent publication) using methods described in class or using newly published statistical methods. Students can either form groups of 2-3 themselves or work individually. A written report (per group) is required and should contain an introduction and description of the data, the biological question of interest, detailed descriptions of the analysis and statistics performed, and a discussion of the results. The report should also include the R code used in the analysis. Detailed instructions about the project and paper will be described in class. A brief project proposal will be due mid-way through the semester (as noted above) and completing this step is part of the final grade. In addition to the report, in-class presentations (5-10 mins) will be scheduled for the last week of class. The exact scheduling details will depend on the size of the class and be announced in advance. The written report is due on the last day of the presentations. Details regarding expectations of the presentation and report will be discussed in class.

Grading

Requirement	% of final grade
Weekly quiz	5%
Assessment 1 (Genetics)	10%
Assessment 2 (Overview)	25%
Problem sets/Participation	15% / 5%
Project Proposal	10%
Project Presentation	15%
Project Paper	15%

Point system used:

Points Earned	93-100	90-92	87-89	83-86	80-82	77-79	73-76	70-72	67-69	63-66	60-62	Below 60
Letter Grade	A	A-	B+	B	B-	C+	C	C-	D+	D	D-	E

Policy Related to Make up work

Full credit will be given for assignments turned in on the due date (by 11:59pm). Assignments turned in the next school day after the due date will have a maximum possible credit of 80%. Assignment turned in two school days after the due date will have a maximum credit of 50%, after this period, late assignments will receive no credit. If you know ahead of time you will be unavailable for an [accepted](#) reason, it is preferred that you turn in the assignment before the due date. Otherwise, you must contact me before the assignment is due with an acceptable reason to avoid penalties for late work.

Please note: Any requests for make-ups due to technical issues MUST be accompanied by the UF Computing help desk (<http://helpdesk.ufl.edu/>) correspondence. You MUST e-mail me (rbacher@ufl.edu) within 24 hours of the technical difficulty.

Policy Related to Required Class Attendance

Attendance is not taken for a grade, but students are expected to be at all class sessions and are responsible for any missed materials. If you know you will be absent, please notify me in advance. Requirements for class attendance and make-up exams, assignments, and other work in this course are consistent with university policies that can be found at:

<https://catalog.ufl.edu/ugrad/current/regulations/info/attendance.aspx>

Student Expectations, Roles, and Opportunities for Input

Expectations Regarding Course Behavior

Students are expected to spend an average of at least 2-1/2 hours per week per credit hour on the course exclusive of class time. This time includes but is not limited to reading, research, preparation for class, and course work. Cell phones should not be used during class. Laptops are permissible and encouraged for note-taking or class related exercises. Questions in class are highly encouraged and should be addressed to the entire class to benefit everyone.

Communication Guidelines

For posting on Canvas or e-mails, please adhere to Netiquette Guidelines: <http://teach.ufl.edu/wp-content/uploads/2012/08/NetiquetteGuideforOnlineCourses.pdf>

Academic Integrity

Students are expected to act in accordance with the University of Florida policy on academic integrity. As a student at the University of Florida, you have committed yourself to uphold the Honor Code, which includes the following pledge:

“We, the members of the University of Florida community, pledge to hold ourselves and our peers to the highest standards of honesty and integrity.”

You are expected to exhibit behavior consistent with this commitment to the UF academic community, and on all work submitted for credit at the University of Florida, the following pledge is either required or implied:

“On my honor, I have neither given nor received unauthorized aid in doing this assignment.”

It is your individual responsibility to know and comply with all university policies and procedures regarding academic integrity and the Student Honor Code. Violations of the Honor Code at the University of Florida will not be tolerated. Violations will be reported to the Dean of Students Office for consideration of disciplinary action. For additional information regarding Academic Integrity, please see Student Conduct and Honor Code or the Graduate Student Website for additional details:

<https://www.dso.ufl.edu/sccr/process/student-conduct-honor-code/>
<http://gradschool.ufl.edu/students/introduction.html>

Please remember cheating, lying, misrepresentation, or plagiarism in any form is unacceptable and inexcusable behavior.

Recording Within the Course:

Students are allowed to record video or audio of class lectures. However, the purposes for which these recordings may be used are strictly controlled. The only allowable purposes are (1) for personal educational use, (2) in connection with a complaint to the university, or (3) as evidence in, or in preparation for, a criminal or civil proceeding. All other purposes are prohibited. Specifically, students may not publish recorded lectures without the written consent of the instructor.

A “class lecture” is an educational presentation intended to inform or teach enrolled students about a particular subject, including any instructor-led discussions that form part of the presentation, and delivered by any instructor hired or appointed by the University, or by a guest instructor, as part of a University of Florida course. A class lecture does not include lab sessions, student presentations, clinical presentations such as patient history, academic exercises involving solely student participation, assessments (quizzes, tests, exams), field trips, private conversations between students in the class or between a student and the faculty or lecturer during a class session.

Publication without permission of the instructor is prohibited. To “publish” means to share, transmit, circulate, distribute, or provide access to a recording, regardless of format or medium, to another person (or persons), including but not limited to another student within the same class section.

Additionally, a recording, or transcript of a recording, is considered published if it is posted on or uploaded to, in whole or in part, any media platform, including but not limited to social media, book, magazine, newspaper, leaflet, or third party note/tutoring services. A student who publishes a recording without written consent may be subject to a civil cause of action instituted by a person injured by the publication and/or discipline under UF Regulation 4.040 Student Honor Code and Student Conduct Code.

Online Faculty Course Evaluation Process

Students are expected to provide professional and respectful feedback on the quality of instruction in this course by completing course evaluations online via GatorEvals. Guidance on how to give feedback in a professional and respectful manner is available at

<https://gatorevals.aa.ufl.edu/students/>. Students will be notified when the evaluation period opens, and can complete evaluations through the email they receive from GatorEvals, in their Canvas course menu under GatorEvals, or via <https://ufl.bluera.com/ufl/>. Summaries of course evaluation results are available to students at <https://gatorevals.aa.ufl.edu/public-results/>.

SUPPORT SERVICES

Accommodations for Students with Disabilities

Students with disabilities who experience learning barriers and would like to request academic accommodations should connect with the Disability Resource Center by visiting <https://disability.ufl.edu/students/get-started/> . It is important for students to share their accommodation letter with their instructor and discuss their access needs, as early as possible in the semester. The College is committed to providing reasonable accommodations to assist students in their coursework.

Counseling and Student Health

Students sometimes experience stress from academic expectations and/or personal and interpersonal issues that may interfere with their academic performance. If you find yourself facing issues that have the potential to or are already negatively affecting your coursework, you are encouraged to talk with an instructor and/or seek help through University resources available to you.

- The Counseling and Wellness Center 352-392-1575 offers a variety of support services such as psychological assessment and intervention and assistance for math and test anxiety. Visit their web site for more information: <http://www.counseling.ufl.edu>. Online and in person assistance is available.
- You Matter We Care website: <http://www.umatter.ufl.edu/>. If you are feeling overwhelmed or stressed, you can reach out for help through the You Matter We Care website, which is staffed by Dean of Students and Counseling Center personnel.
- The Student Health Care Center at Shands is a satellite clinic of the main Student Health Care Center located on Fletcher Drive on campus. Student Health at Shands offers a variety of clinical services. The clinic is located on the second floor of the Dental Tower in the Health Science Center. For more information, contact the clinic at 392-0627 or check out the web site at: <https://shcc.ufl.edu/>
- Crisis intervention is always available 24/7 from:
Alachua County Crisis Center: (352) 264-6789
<http://www.alachuacounty.us/DEPTS/CSS/CRISISCENTER/Pages/CrisisCenter.aspx>

Do not wait until you reach a crisis to come in and talk with us. We have helped many students through stressful situations impacting their academic performance. You are not alone so do not be afraid to ask for assistance.