

University of Florida
College of Public Health & Health Professions Syllabus
PHC 7594 GENETIC EPIDEMIOLOGY (3 CREDIT HOURS)

Summer A: 2024

Monday and Wednesday: 2:00pm – 4:45pm

Delivery Format: On-Campus

Course Website or E-Learning <https://ufl.instructure.com>

Instructor Name: Jinying Zhao, MD, PhD.

Room Number: Department of Epidemiology, Room 4240C

Phone Number: (352) 273-5933

Email: jzhao66@ufl.edu

Office hours: By appointment

Teaching Assistant: Mingjing Chen (mingjingchen@ufl.edu)

Preferred Course Communications: email

Prerequisites: PHC 6000 (Epi Methods I), PHC 6011 (Epi Methods II, can be taken concurrently), and PHC 6050c (Biostat Methods I). Talk to the instructor for a prerequisite waiver or further information. Course is designed for master level (MS/MPH) or PhD students.

Audience for this course

- Students who are interested in genetic epidemiology of human disease
- Students who will be working with genetic data
- Students who are interested in statistical analysis of genetic data
- Students who are interested in learning research methods in genetics, and those who want to expand their knowledge in genetics and statistical genetics.

PURPOSE AND OUTCOME

Course Overview

This course covers fundamental concepts, terminologies and principles in genetic epidemiology. Study designs and analytical methods for gene mapping will be covered. Students will also gain hands-on experience in genetic linkage and association analysis in computer lab. At the completion of this course, students should be able to critically review and discuss genetic epidemiologic literatures, provide input on the design of genetic epidemiologic studies, identify and apply appropriate tests for genetic analysis, and interpret results of genetic linkage and association analysis. This course is one of the available advanced epidemiology methods courses for the PhD program.

Relation to Program Outcomes

The main purposes of this 3-credit hour course are to introduce and consolidate: (1) basic concepts, terminologies, and theories in human population genetics and genetic epidemiology; (2) study design of genetic epidemiological research; (3) necessary analytical skills in genetic epidemiology and precision health. This course will also teach students how to design an efficient genome-wide linkage and association studies and how to interpret research findings of genetic epidemiological studies in large-scale human populations.

Course Objectives and/or Goals

- Describe basic concepts and terminologies in human population genetics
- Explain fundamental principles and theories in genetic epidemiology
- Describe various genetic markers commonly used in genetic epidemiology studies
- Conduct Hardy-Weinberg Equilibrium (HWE) test and calculate allele and genotype frequencies
- Calculate test statistics for genetic analysis, such as LOD scores, IBD and IBS, etc
- Compare and contrast genetic linkage and association analysis
- Compare and contrast family-based and population-based studies
- Interpret results of genetic linkage and association analyses, including genome-wide linkage and genome-wide association studies (GWAS)
- Describe HapMap and explain tagging SNPs
- Discuss issues related to human genetic studies, such as genetic heterogeneity, population admixture and multiple comparisons

Instructional Methods

Since genetic epidemiology is one of the fastest growing fields, there is no one textbook that covers all the topics or follows up the progresses. The format of this course will be a combination of the following:

- Didactic lectures: The course will consist of lectures to introduce fundamental concepts, terminologies, and principles in the areas of human population genetics and genetic epidemiology.
- Homework: Students will be responsible for completing the assigned homework and handing in their homework on time.
- Assigned readings: Students are responsible for completing the assigned readings and should be prepared to discuss each reading assignment in class.
- In-class exercises: The course will include in-class exercises to help students understand the key concepts and theories discussed in class.
- Examination: There will be one closed book in-class final exam at the end of this course.
- Oral presentation: Students are required to orally present a chosen topic in the area of genetic epidemiology. Students will work as groups (3-4 students in each group, 15-20 minutes for each group) to present their work in PowerPoint slides. Journal articles for presentation can be selected based on the students' interests, but prior approval from the course instructor is required in order to avoid repeating of the same or similar topic.

DESCRIPTION OF COURSE CONTENT

Course Schedule: This represents a schedule for summer 2024. Schedule changes will be announced in advance.

Date	Class	Topic	HW	HW due	Readings
5/13/24	Lecture 1	<ul style="list-style-type: none"> Course introduction Introduction to genetic epidemiology Key concepts in human genetics 	HW1: Basic concepts in hum genet		<u>Reading 1</u> : Introduction to genet epi
5/15/24	Lecture 2	<ul style="list-style-type: none"> HWE and its applications 	HW 2: HWE		
5/20/24	Lecture 3	<ul style="list-style-type: none"> Linkage disequilibrium (LD) Principles of Mendelian inheritance 	HW 3: Genotype and allele frequency	HW 1 due	
5/22/24	Lecture 4	<ul style="list-style-type: none"> Familial aggregation Heritability analysis Segregation analysis 	HW 4: pedigree, concordance/discordance	HW 2 due	<u>Reading 2</u> : Familial aggregation and heritability of pyloric stenosis <u>Reading 3</u> : Segregation for early onset major depression
5/27/24	Memorial day	No class			
5/29/24	Lecture 5	<ul style="list-style-type: none"> Genetic markers TagSNPs & Haplotype blocks 	HW 5: genetic marker, heterozygosity	HW 3 due	<u>Reading 4</u> : Genetic markers <u>Reading 5</u> : TagSNPs & haplotype block
6/3/24	Lecture 6	<ul style="list-style-type: none"> Parametric linkage Nonparametric linkage 		HW 4 due	<u>Reading 6</u> : Linkage for prostate cancer <u>Reading 7</u> : Linkage for essential hypertension
6/5/24	Lecture 7	<ul style="list-style-type: none"> Family-based association studies Case-control association studies GWAS and issues related to genetic associated studies 		HW 5 due	<u>Reading 8</u> : Family-based GWAS <u>Reading 9</u> : GWAS for lung cancer
6/10/24		<ul style="list-style-type: none"> Course summary and review 			
6/12/24		<ul style="list-style-type: none"> Students oral presentation 			
6/17/24		<ul style="list-style-type: none"> Prepare for final 			
6/19/24		<ul style="list-style-type: none"> Final exam (closed book, in-class exam) 			

Course Textbook (recommended but not required)

- *Statistical Methods in Genetic Epidemiology* by Duncan Thomas Publisher: Oxford University Press. ISBN: 978-0195159394
- Useful reference books:
 - *Principles of Population Genetics*, 4rd Edition, Daniel Hartl & Andrew Clark, 2007, ISBN 978 0 87893 308 2
 - *An introduction to Genetic Epidemiology*. Edited by Lyle J. Palmer, Paul R. Burton and George D. Smith. The Policy Press 2011. ISBN 978 1 86134 897 5
 - *Genetics in Medicine*, Nussbaum Robert L, et al. ISBN: 978 1 41603 080 5
- Specific readings related to each topic will change from year to year based on the latest publications.

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

- helpdesk@ufl.edu
- (352) 392-HELP - select option 2
- <https://helpdesk.ufl.edu/>

Additional Academic Resources

[Career Connections Center](#): Reitz Union Suite 1300, 352-392-1601. Career assistance and counseling services.

[Library Support](#): Various ways to receive assistance with respect to using the libraries or finding resources.

[Teaching Center](#): Broward Hall, 352-392-2010 or to make an appointment 352- 392-6420. General study skills and tutoring.

[Writing Studio](#): 2215 Turlington Hall, 352-846-1138. Help brainstorming, formatting, and writing papers.

Student Complaints On-Campus: [Visit the Student Honor Code and Student Conduct Code webpage for more information.](#)

On-Line Students Complaints: [View the Distance Learning Student Complaint Process.](#)

Course Requirements

- Attend the class and participate in group discussions

- Complete and return the assigned homework
- Complete the assigned reading and be prepared to discuss in class
- Complete one in-class written exam

ACADEMIC REQUIREMENTS AND GRADING

Assignments

- There will be 5 homework assignments for this class. The homework will be due one week after the assignment is released. You can hand in a hard copy or email your homework to the TA before deadline. Late homework will not be accepted except for emergencies.
- There will also be 9 assigned readings of literature articles relevant to the topics in genetic epidemiology. Students are required to read the articles and be prepared to discuss them in class.

Grading

The final grade will be based on the following components: 5 assigned homework, class participation, oral presentation, and final exam.

- Homework (50%): You will have 5 assignments, each worth 10 points. Each assignment will be graded based on whether the calculation is correct or not. Data interpretation will also count towards to the score of each assignment.
- Class participation (10%): Students are required to attend all class sessions and to actively participate in classroom discussions on the assigned reading or other related topics. If a student is more than 20 minutes late to the class, the student will be treated as not present.
- Oral presentation (20%): Students are required to orally present a topic related to genetic epidemiology. The oral presentation will be graded based on several factors including how well the student understands the chosen topic, interpretation of the results, answers to questions, etc. they The topic can be chosen by students, but prior approval is required from the instructor to avoid repeating of the same or similar topics.
- Final exam (20%): There will be one in-class, closed book exam. Any topic covered in the class will possibly be included in the exam (unless specifically stated by the instructor)

Point system used (i.e., how do course points translate into letter grades).

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

Please be aware that a C- is not an acceptable grade for graduate students. The GPA for graduate students must be 3.0 based on 5000 level courses and above to graduate. A grade of C counts toward a

graduate degree only if based on credits in courses numbered 5000 or higher that have been earned with a B+ or higher.

Letter Grade	Grade Points
A	4.0
A-	3.67
B+	3.33
B	3.0
B-	2.67
C+	2.33
C	2.0
C-	1.67
D+	1.33
D	1.0
D-	0.67
E	0.0
WF	0.0
I	0.0
NG	0.0
S-U	0.0

More information on UF grading policy may be found at:

<http://gradcatalog.ufl.edu/content.php?catoid=10&navoid=2020#grades>

Exam Policy

If you cannot submit an assignment on time, it is your responsibility to communicate that information ahead of time to the instructor. Late work will be accepted on a case-by-case basis.

Policy Related to Make up Exams or Other Work

Please note: Any requests for make-ups due to technical issues MUST be accompanied by the ticket number received from LSS when the problem was reported to them. The ticket number will document the time and date of the problem. You MUST e-mail the instructor within 24 hours of the technical difficulty if you wish to request a make-up.

Policy Related to Required Class Attendance

Students are expected to attend all class sessions and participate in classroom discussions. If a student is more than 20 minutes late to the class, the student will be treated as not present, and will not receive any points toward their class participation grade for that day. Students who cannot attend a class should

inform the instructor via email prior to the date of the class, or on the day of the absence for illness or emergency.

Excused absences must be consistent with university policies in the Graduate Catalog (<https://catalog.ufl.edu/graduate/regulations/#text>). Additional information can be found here: <https://catalog.ufl.edu/ugrad/current/regulations/info/attendance.aspx>

STUDENT EXPECTATIONS, ROLES, AND OPPORTUNITIES FOR INPUT

Expectations Regarding Course Behavior

- Attend the class and participate in group discussions
- Complete and return the assigned homework
- Complete the assigned reading and be prepared to discuss in class
- Complete one in-class written final exam. No internet and electronics (laptops, phones, recording devices, etc.) are allowed during the exam.
- Complete an oral presentation with your group

Communication Guidelines

Email correspondence and all interactions on e-Learning should follow the etiquette of business emails (see UF's *Netiquette Guide for Online Courses* for guidance at <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.

Policy Related to Guests Attending Class:

Only registered students are permitted to attend class. However, we recognize that students who are caretakers may face occasional unexpected challenges creating attendance barriers. Therefore, by exception, a department chair or his or her designee (e.g., instructors) may grant a student permission to bring a guest(s) for a total of two class sessions per semester. This is two sessions total across all courses. No further extensions will be granted. Please note that guests are **not** permitted to attend either cadaver or wet labs. Students are responsible for course material regardless of attendance. For additional information, please review the Classroom Guests of Students policy in its entirety. Link to full policy: <https://phhp.ufl.edu/policy-classroom-guests-of-students/>

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

If you require classroom accommodation because of a disability, it is strongly recommended you register with the Dean of Students Office <http://www.dso.ufl.edu> within the first week of class or as soon as you believe you might be eligible for accommodations. The Dean of Students Office will provide documentation of accommodations to you, which you must then give to me as the instructor of the course to receive accommodations. Please do this as soon as possible after you receive the letter. Students with disabilities should follow this procedure 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>. On line and in person assistance is available.
- **U 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>
- **University Police Department:** [Visit UF Police Department website](#) or call 352-392-1111 (or 9-1-1 for emergencies).
- **UF Health Shands Emergency Room / Trauma Center:** For immediate medical care call 352-733-0111 or go to the emergency room at 1515 SW Archer Road, Gainesville, FL 32608; [Visit the UF Health Emergency Room and Trauma Center website](#).

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.

Inclusive Learning Environment

Public health and health professions are based on the belief in human dignity and on respect for the individual. As we share our personal beliefs inside or outside of the classroom, it is always with the understanding that we value and respect diversity of background, experience, and opinion, where every individual feels valued. We believe in, and promote, openness and tolerance of differences in ethnicity and culture, and we respect differing personal, spiritual, religious and political values. We further believe that celebrating such diversity enriches the quality of the educational experiences we provide our students and enhances our own personal and professional relationships. We embrace The University of Florida's Non-Discrimination Policy, which reads, "The University shall actively promote equal opportunity policies and practices conforming to laws against discrimination. The University is committed to non-discrimination with respect to race, creed, color, religion, age, disability, sex, sexual orientation, gender identity and expression, marital status, national origin, political opinions or affiliations, genetic information and veteran status as protected under the Vietnam Era Veterans' Readjustment Assistance Act." If you have questions or concerns about your rights and responsibilities for inclusive learning environment, please see your instructor or refer to the Office of Multicultural & Diversity Affairs website: www.multicultural.ufl.edu.