

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

Summer A: 2020

Monday and Wednesday: 9:00am – 12:45pm

Delivery Format: Asynchronous

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

Instructor Name: Jinying Zhao, MD, PhD

Department of Epidemiology

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Preferred course communications: email or by appointment

**Prerequisites:** PHC 6000 (Epi Methods I), PHC 6011 (Epi Methods II, can be taken concurrently), and PHC 6050c (Biostatistical Methods I). Talk to the instructor for a prerequisite waiver or further information. The course is designed for PhD students. MS/MPH students may contact the instructor for permission.

#### **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 and principles in genetic epidemiology. At the completion of this course, students are expected to critically discuss literature, design and conduct basic genetic analysis, and interpret research findings. This course is one of the available advanced epidemiology methods courses for the PhD program.

##### **Relation to Program Outcomes**

This course covers key concepts and theories in human population genetics and genetic epidemiology. This course will help graduate students to achieve PhD in Epidemiology Program Student Learning Outcome #2: Prepare to become an independent researcher in the field of

Epidemiology and PhD in Epidemiology Program Student Learning Outcome #3: Illustrate a thorough understanding of epidemiology concepts.

### **Course Objectives and/or Goals**

- Learn 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**

Because 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 covering key concepts 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 apply key concepts and theories discussed in class to address genetic epidemiological questions.
- Examination: There will be one closed book in-class final exam at the end of this course.
- Computer lab: Student will gain hands-on experience in genetic data analyses in computer lab.
- 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, 20-25 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**

**Outline/Course Schedule:** This represents a schedule for summer 2018. Schedule changes will be announced in advance.

Class	Topic	HW	HW due	Readings
Week 1	<ul style="list-style-type: none"> <li>Course introduction</li> <li>Key concepts in genetic epidemiology</li> <li>Key concepts in human genetics</li> </ul>	HW 1: Basic concepts in genetics		<u>Assigned reading #1:</u> Genetic epidemiology and public health
Week 1	<ul style="list-style-type: none"> <li>Population genetics I</li> <li>Population genetics II</li> </ul>	HW 2: HWE		
Week 2	<ul style="list-style-type: none"> <li>Population genetics III</li> <li>Principles of Mendelian inheritance</li> </ul>	HW 3: genotype freq	HW 1 due	
Week 2	<ul style="list-style-type: none"> <li>Familial aggregation and heritability analysis</li> <li>Segregation analysis</li> </ul>	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
Week 3	Memorial Day – No class			
Week 3	<ul style="list-style-type: none"> <li>Genetic markers</li> <li>TagSNPs &amp; Haplotype blocks</li> <li>Computer lab I – HapMap and TagSNP selection</li> </ul>	HW 5: genetic marker, heterozygosity	HW 3 due	<u>Reading 4:</u> Genetic markers <u>Reading 5:</u> TagSNPs & haplotype block
Week 4	<ul style="list-style-type: none"> <li>Parametric linkage analysis</li> <li>Nonparametric linkage analysis</li> </ul>		HW 4 due	<u>Reading 6:</u> Linkage for prostate cancer <u>Reading 7:</u> Linkage for essential hypertension
Week 4	<ul style="list-style-type: none"> <li>Family-based association studies</li> <li>Population-based association studies</li> <li>GWAS and issues related to genetic associated studies</li> </ul>		HW 5 due	<u>Reading 8:</u> GWAS (family-based design) <u>Reading 9:</u> GWAS (case-control design)
Week 5	<ul style="list-style-type: none"> <li>Computer lab II – Genetic linkage and association analysis</li> </ul>			
Week 5	Students oral presentation			
Week 6	<ul style="list-style-type: none"> <li>Course review &amp; summary</li> <li>Introduction to Epigenetics (if time allows)</li> </ul>			
Week 6	Last Day of Class – 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:

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

### **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

### **Method of Evaluation and Grading**

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

- Homework (50%): There will be five homework assignments for this class; each will count 12 points towards the percentage total for homework assignments. Homework assignments will be composed of questions related to the genetic epidemiology methods and calculations taught in the weekly lectures (e.g., Hardy-Weinberg Equilibrium (HWE) test, allele and genotype frequencies, LOD scores, IBD and IBS, etc.) and to the interpretation of genetic epidemiological data (e.g., results of genetic linkage and association analyses, including genome-wide linkage and genome-wide association studies). The homework assignments will give students practice working with genetic epidemiological data and will prepare students for their oral presentation and their final exam. You can hand in a hard copy or email your homework to the TA before deadline. Late homework will not be accepted except for emergency situations. Each assignment will be graded according to the below rubric.
- 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; thus they will forfeit 1/12<sup>th</sup> of 10% of their grade for each class session.
- 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. 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)

Homework Grading Rubric				
	Points assigned			
Criteria	4 points	2points	0 point	Total points possible
Logical thought process in working problems	Problem solution attacked logically and systematically	Logic present for some steps in the calculation; logic not based in module resources	No clear logic shown for any steps in the calculation	4
Calculation correct	Calculation correct	Some steps in calculation correct	No work shown; inaccurate answer	4
Correct interpretation of data	Interpretation correct	Interpreted some of the calculations correctly	No calculations interpreted correctly	4
<b>Total</b>				<b>12</b>

This course will be graded following the policies described here

<http://gradcatalog.ufl.edu/content.php?catoid=4&navoid=907&hl=grades&returnto=search#grades>

<b>Points earned</b>	93-100	90-92	87-89	83-86	80-82	77-79	73-76	70-72	67-69	63-66	60-62	<60
<b>Letter Grade</b>	A	A-	B+	B	B-	C+	C	C-	D+	D	D-	E
<b>Grade points</b>	4.0	3.67	3.33	3.0	2.67	2.33	2.0	1.67	1.33	1.0	0.67	0.0

For details on the meaning of letter grades and university policies related to them, please see the Registrar's Grade Policy regulations at: <http://catalog.ufl.edu/ugrad/current/regulations/info/grades.aspx>

### Policy Related to Make up Exams or Other Work

If you cannot submit any assignment on time, it is your job to communicate that information ahead of time to the instructor.

Make-up work will be allowed with permission of the course instructor on an individual basis after an excused absence. Please consult the university guidelines for more information on makeup policies: <https://catalog.ufl.edu/ugrad/current/regulations/info/attendance.aspx>.

Please note: Any requests for make-ups due to technical issues MUST be accompanied by the ticket number received from the Help Desk 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. Please note all faculty are bound by the UF policy for excused absences. For information regarding the UF Attendance Policy see the Registrar website for additional details:

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

### **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:

<http://facstaff.phhp.ufl.edu/services/resourceguide/getstarted.htm>

## **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.

### **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.

### **Citations and Plagiarism**

The two key purposes of citation are to: 1) give appropriate credit to the authors of information, research findings, and/or ideas (and avoid plagiarism), and 2) facilitate access by your readers to the sources you use in your research.

**Quotations:** When directly quoting an outside source, the borrowed text, regardless of the amount, must be surrounded by quotation marks or block quoted. Quoted text over two lines in length should be single-spaced and indented beyond the normal margins. Every quote must include a source—the author, title, volume, page numbers, etc.—whether an internal reference, footnote, or endnote is used in conjunction with a bibliography page.

**Paraphrasing or Citing an Idea:** When summarizing an outside source in your own words or citing another person’s ideas, quotation marks are not necessary, but the source must be included. This includes, but is not confined to, personal communications from other students, faculty members, experts in the field, summarized ideas from published or unpublished resource, and primary methods derived from published or unpublished sources. Use the general concept of “when in doubt – cite.”

Plagiarism is a serious violation of the academic honesty policy of the College. If a student plagiarizes others’ material or ideas, UF Policies on Honesty and honor code violations, noted above, will be followed.

Generally speaking, the three keys of acceptable citation practice are: 1) thoroughness, 2) accuracy, and 3) consistency. In other words, be sure to fully cite all sources used (thoroughness), be accurate in the citation information provided, and be consistent in the citation style you adopt. All references should include the following elements: 1) last names along with first and middle initials; 2) full title of reference; 3) name of journal or book; 4) publication city, publisher, volume, and date; and 5) page numbers referenced. When citing information from the Internet, include the WWW address at the end, with the “access date” (i.e., when you obtained the information), just as you would list the document number and date for all public documents. When citing ideas or words from an individual that are not published, you can write “personal communication” along with the person’s name and date of communication.

### **Use of unauthorized assistance resources**

As a graduate student at UF, you are expected to present your own work for grading. Unauthorized sources of help, including commercially available software and services are not allowed. Even though the students will not be graded on their grammar, it is expected that as graduate students you will have sufficient English language skills to convey your thoughts in an organized and understandable manner. If the assignment is unreadable, it will not be graded and will be assigned zero points. Use of unauthorized assistance sources is not permitted and represents a violation of the Student Honor Code. Violations will be reported to the Dean of Students Office and appropriate penalties/sanctions will be determined. If

English is your Second Language, you may visit the UF Writing Program Website to learn about available help.

### **Online Faculty Course Evaluation Process**

Students are expected to provide feedback on the quality of instruction in this course by completing online evaluations at <https://evaluations.ufl.edu>. Evaluations are typically open during the last two or three weeks of the semester, but students will be given specific times when they are open. Summary results of these assessments are available to students at <https://evaluations.ufl.edu/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.
- 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.