

University of Florida
College of Public Health & Health Professions Syllabus
PHC 6937 GENETIC EPIDEMIOLOGY (3 CREDIT HOURS)

Summer section A: 2017

Monday and Wednesday: 9:00am – 12:30pm

Delivery Format: On-Campus

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

Instructor Name: Jinying Zhao, MD, PhD

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Office hours: By appointment

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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. 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

Genetic epidemiology is a rapidly evolving field of epidemiologic research that utilizes highly specialized molecular and statistical methods to identify genetic factors that might be involved in disease etiology. This introductory course will cover fundamental concepts, terminologies and principles in human population genetics and molecular biology relevant to understanding approaches in genetic epidemiology, including allele, genotype, haplotype, models of inheritance, Hardy-Weinberg equilibrium (HWE), linkage disequilibrium (LD), and population stratification. Study designs and analytical methods for genetic epidemiological studies of human disease in families and unrelated individuals, including genetic linkage (both parametric and non-parametric linkage) and association analyses (both candidate gene-based and GWAS) will be discussed. Issues related to genetic studies, such as genetic heterogeneity, population stratification and multiple testing will also be covered. Examples relevant to public health will be emphasized, including the application of these important fields to studies of human common

chronic diseases such as cardiovascular disease, type 2 diabetes, obesity and cancer. Students will also have chance to conduct genetic linkage and association analysis in the computer lab, participate in group discussions and critical review of journal articles relevant to genetic epidemiology. There will be assigned homework and readings, in-class written exam and oral presentation for chosen diseases.

The course will provide students with a focused exposure to major concepts and theories in genetic epidemiology for human diseases. 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 for both qualitative and quantitative outcomes using either families or unrelated individuals, and interpret the results of genetic linkage and association analysis.

Course Objectives

- 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
- Explain the difference between genetic linkage and association analysis
- Explain the difference between 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
- Provide hands-on, practical experiences in tagSNP selection, genetic linkage and association analysis

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 mainly 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 hand in their homework before deadline.
- **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 student 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.
- **Computer lab:** Student will have chance to perform genetic linkage and association

analyses using genetic software in the 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, 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

Outline/Course Schedule: This represents a likely topic schedule for summer 2017. Schedule changes will be announced in advance.

Class	Topic	HW	HW due	Readings
Week 1 05/08/2017	<ul style="list-style-type: none"> • Course introduction • Introduction to genetic epidemiology • Key concepts in human genetics 	HW 1		<u>Reading 1</u> : Introduction to genetic epidemiology
Week 1 05/10/2017	<ul style="list-style-type: none"> • Population genetics I • Population genetics II 	HW 2		
Week 2 05/15/2017	<ul style="list-style-type: none"> • Population genetics III • Principles of Mendelian inheritance 	HW 3	HW 1 due	
Week 2 05/17/2017	<ul style="list-style-type: none"> • Familial aggregation and heritability analysis • Segregation analysis 	HW 4	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 05/22/2017	<ul style="list-style-type: none"> • Genetic markers • TagSNPs & Haplotype blocks • Computer lab I – HapMap and TagSNP selection 	HW 5	HW 3 due	<u>Reading 4</u> : Genetic markers <u>Reading 5</u> : TagSNPs & haplotype block
Week 3 05/24/2017	<ul style="list-style-type: none"> • Parametric linkage analysis • Nonparametric linkage analysis 		HW 4 due	<u>Reading 6</u> : Linkage for prostate cancer <u>Reading 7</u> : Linkage for essential hypertension
Week 4 05/29/2017	Memorial Day – No class			
Week 4 05/31/2017	<ul style="list-style-type: none"> • Family-based association studies • Population-based 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
Week 5 06/05/2017	<ul style="list-style-type: none"> • Computer lab II – Genetic linkage and association analysis 			
Week 5 06/07/2017	Students oral presentation			
Week 6 06/12/2017	<ul style="list-style-type: none"> • Course review & summary • Introduction to Epigenetics (if time allows) 			
Week 6 06/14/2017	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

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 the two in-class written examinations: midterm and final 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 assigned homework for this class. Late homework will not be accepted except for emergency situations.
- 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 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%): This will be an in-class, closed book exam. Any topic covered in the class will possibly be included in the exam (unless specifically stated by the instructor)

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>

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
Grade points	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 greater detail on the meaning of letter grades and university policies related to them, 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

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>.

Policy Related to Required Class Attendance

Students are expected to attend all class sessions and participate in classroom discussions. 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>

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 the two in-class written examinations: midterm and final exam

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 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 will result in zero points on the written assignment and a report to the Dean of Student's Office. 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, you must register with the Dean of Students Office <http://www.dso.ufl.edu> within the first week of class. The Dean of Students Office will provide documentation of accommodations to you, which you then give to me as the instructor of the course to receive accommodations. Please make sure you provide this letter to me by the end of the second week of the course. 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.