

University of Florida
College of Public Health & Health Professions Syllabus
PHC 6598: Foundations in Precision Medicine: Genetic Epidemiology (1 credit hour)
 Spring 2020
 Delivery Format: Online
 Canvas or E-Learning: <https://ufl.instructure.com/courses/387209>

Instructor Name: **Alexander Parker PhD, MS**

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Teaching Assistant: **Abdelrahman H. Elsayed, M.Sc.**, Graduate Research Assistant, aelsayed@ufl.edu

Preferred Course Communications: Canvas mail, discussion board, or by appointment

Prerequisites

Enrollment in the Certificate for Precision Medicine or permission of the instructor.

PHC 6598: Genetic Epidemiology is one of the foundational courses for the Certification in Precision Medicine. It follows to other foundational courses. Medical Molecular Genetics and Genomic Technologies, which contain information on molecular genetics and genome analysis that will be assumed to have been mastered prior to this course which will focus on their application to genetic epidemiology.

PURPOSE AND OUTCOME

Course Overview

Genetic epidemiology, a rapidly evolving field of research, utilizes specialized molecular and statistical methods to identify associations between genetic factors and phenotypes that may causally related to human disease. This course provides an exposure to fundamental concepts, terminologies and principles in human population genetics and applied molecular biology relevant to design, conduct and interpretation of genetic epidemiologic studies.

Relation to Program Outcomes

This 1-credit course is a required, foundational course for the Precision Medicine Graduate Certificate Program, and covers the following overarching program competencies:

- Assess patient-specific and epidemiologic data to determine clinical utility and practical use of genomic or pharmacogenomics tests to established association with phenotypes of interest in clinical purpose and/or population health
- Apply current and emerging development in genetics, genomics, pharmacogenomics and clinical informatics to integrate genomic medicine into practice.

Course Objectives and/or Goals

- Describe basic concepts and terminologies in human population genetics
- Explain fundamental principles and theories in genetic epidemiology
- Review design and interpretation of studies of familial aggregation of disease
- Describe various genetic markers commonly used in genetic epidemiology studies
- Explain the difference between genetic linkage and association analyses
- Interpret results of studies using candidate gene approaches results of studies of genome-wide
- Interpret (agnostic) approaches
- Understand the types of bias that could occur in genetic epidemiology studies
- Critically review articles in the area of genetic epidemiology
- Describe the application of genetic epidemiology, study results to other fields of research and healthcare

DESCRIPTION OF COURSE CONTENT
Topical Outline/Course Schedule

Week		Topics	Contact Hrs.
Week 1 (3/16-3/22)	Lecture	Course Introduction	1.0
	Lecture	Introduction to Genetic Epidemiology, Family History, Family Studies, Heritability	1.0
	Assignment	Assignment #1 Due	1.0
	Quiz	Quiz #1	.5
	Assignment	Asynchronous Discussion Board Participation	1.0
	Readings	<p>Required Textbook: Burton PR, Tobin MD, Hopper JL. Key Concepts in Genetic Epidemiology; Chapter 1 in An Introduction to Genetic Epidemiology (Palmer, Burton, Davey Smith) 2011. (.5 Contact hr.)</p> <p>Journal Articles: Fallin, MD, Duggal P, Beaty TH. Genetic Epidemiology and Public Health: The Evolution for Theory to Technology; American Journal of Epidemiology. 2016; https://doi.org/10.1093/aje/kww001 (.5 Contact hr.)</p>	
Week 2 (3/23-3/29)	Lecture	Genetic Markers and Linkage Studies	.75
	Assignment	Assignment#2 Due	1.0
	Assignment	Asynchronous Discussion Board Participation	1.0
	Quiz	Quiz #2	.5
	Readings	<p>Required Textbook: Teare DM, Barrett JH. Genetic Linkage Studies. Chapter 2 in An introduction to Genetic Epidemiology (Palmer, Burton, Davey Smith) 2011. Journal Articles: International HapMap Consortium. A second generation human haplotype map of over 3.1 million SNPs. Nature 2007; 449: 851-861 (.5 Contact Hours)</p>	
Week 3	Lecture	Candidate Genes and Association Studies	.75
	Assignment	Assignment#3 Due	1.0
	Assignment	Asynchronous Discussion Board Participation	1.0
	Quiz	Quiz #3	.5
	Readings	<p>Required Textbook: Cardell HJ, Clayton DG. Genetic Association Studies. Chapter 3 in An Introduction to Genetic Epidemiology (Palmer, Burton, Davey Smith) 2011.</p> <p>Hattersley AT, McCarthy MI. A Question of Standards: What Makes a Good Genetic Association Study? Chapter 5 in An Introduction to Genetic Epidemiology (Palmer, Burton, Davey Smith) 2011.</p>	
Week 4	Lecture	Genome Wide Association Studies	1.0
	Assignment	Assignment#4 Due	1.0
	Assignment	Asynchronous Discussion Board Participation	1.0
	Quiz	Quiz #4	.5
	Readings	<p>Required Textbook: Palmer LJ, Timpson NJ, Evans DM, Davey Smith G, Cardon LR. Mapping Complex Disease Genes Using Linkage Disequilibrium and Genome-wide Association Scans. Chapter 4 in An Introduction to Genetic Epidemiology (Palmer, Burton, Davey Smith) 2011. Journal Articles: Pearson TA, Manolio TA. How to Interpret a Genome-wide Association Study. JAMA 2008; 299:1335-1344 (.75 Contact Hours)</p>	
Week 5	Lecture	Interpretation and Application of genetic Epidemiology	1hr.
	Assignment	Assignment#5 Due	1.0
	Assignment	Asynchronous Discussion Board Participation	1.0
	Quiz	Quiz #5	.5hr.

Readings	<p>Required Textbook: Davey Smith G, Ebrahim S, Lewis S, Palmer LJ; Population Health Aspects of Genetics Epidemiology: Genomic Profiling, Personalized Medicine, and Mendelian Randomization. Chapter 1 in An Introduction to Genetic Epidemiology (Palmer, Burton, Davey Smith) 2011.</p> <p>Journal Articles: Gonzaga- Jauregui C, Lupski JR, Gibbs RA. Human Genome Sequencing in Health and Disease. Annual Review of Medicine 2012; 63:35-61.</p> <p>(1.0 Contact Hours)</p>	
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Instructional Methods

This is an online course that includes viewing of recorded lectures, completion of written assignments, participation in an asynchronous discussion board, and self-review with weekly quizzes. Students will independently view recorded lectures and complete assigned readings on the Canvas website. Then, the student will complete an online quiz on Canvas to assess understanding of the material and also complete an assignment, to be submitted on Canvas. Following the deadline for submitting the assignments, the instructor will grade the assignments, post the grades on Canvas, and then post additional information on a Canvas discussion board thread to assist students in further understanding the course materials. Students are then expected to contribute to the discussion/questions posed by the instructor by replying to the Canvas thread. Throughout all of these learning activities, the instructor and teaching assistant are available to clarify information via Canvas discussion board postings.

Course Materials and Technology

Journal articles, class notes developed by the instructor, or other required reading/resources will be provided to students through the course website.

A relatively concise and inexpensive textbook will be required for the course:

- 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

Two other text books can be recommended but are not required:

- Principles of Population Genetics, 4th Edition, Daniel Hartl & Andrew Clark, 2007, ISBN 978 0 87893 308 2
- Genetics in Medicine, Nussbaum Robert L, et al. ISBN: 978 1 41603 080 5

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

Requirement	Due Date	Total Points	Percentage of Final Grade
Assignments (N=5)	Due each week on Day 7	200 each	50%
On-line Quizzes (N=5)	Due each week on Day 7	100 each	25%
Discussion Board Participation (N=5)	Due each week on Day 7	100 each	25%
			100%

Assignments Grading

Homework Assignments. Each week students will be given a different set of population genetics specific problems to work through, related to the current weeks' topics. Students will submit their assignment responses through the on-line course software (Canvas). Assignments will be graded on the basis of responsiveness to the assignment, completeness and accuracy of response, and quality of writing or documentation of calculations. Further information and grading criteria will be provided in class.

Quizzes will be graded on the basis of correct answers with a score of 100 possible.

Discussion Board. This investigator facilitated discussion board will require the student to create one posting and at least two replies each week. **Postings** will be graded using this rubric:

	Excellent	Good	Fair	Poor	Total
Responsiveness to discussion assignment prompt 50 points	All components of discussion prompt addressed in initial posting 50 – 46 points	Up to 90% of the prompt components addressed in one or more postings 45 – 40 points	Up to 75% of the prompt components addressed in one or more postings 39-35 points	50% or less of the prompt addressed in one or more postings 34 points to 0 points	/50
Application of assigned reading(s) 25 points	Very clear that readings were understood by incorporation into postings 25 – 24 points	Readings were understood and incorporated 23 – 21 points	Somewhat clear that readings were understood by incorporation into postings 20 – 18 points	Unclear that readings were understood by incorporation into postings 17 or less points	/25
Responsiveness to fellow students' posting 25 points	Responsiveness to group discussion and at least two additional replies with multiple points from multiple participants clearly built upon/refuted in postings 25 – 24 points	At least one point from multiple participants clearly built upon/refuted in postings 23 – 21 points	One or more points from one or more participants only vaguely built upon/refuted in postings 20 – 18 points	No evidence that any other postings have been read/Unwitting repetition of questions or points made by others 17 or less points	/25
Total points/100					/100

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

Points earned	90.0 – 100.0	87.0 – 89.9	84.0 – 86.9	80.0 – 83.9	75.0 – 79.9	72.0 – 74.9	69.0 – 71.9	66.0 – 68.9	63.0 – 65.9	60.0 – 62.9	58.0 – 59.9	0 – 57.9
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. A grade of C counts toward a graduate degree only if an equal number of credits in courses numbered 5000 or higher have been earned with an A.

You must include the letter grade to grade point conversion table below. Letter grade to grade point conversions are fixed by UF and cannot be changed.

Letter Grade	A	A-	B+	B	B-	C+	C	C-	D+	D	D-	E	WF	I	NG	S- U
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	0.0	0.0	0.0	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://gradcatalog.ufl.edu/content.php?catoid=10&navoid=2020#grades>

<http://catalog.ufl.edu/ugrad/current/regulations/info/grades.aspx>

Policy Related to Make up Exams or Other Work

Students will be allowed to make-up quizzes, assignments, and discussion board activities for acceptable reasons as described in the Graduate Catalog. See:

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

If you are unable to submit an assignment on time, it is your responsibility to contact the instructor through Canvas mail as soon as possible to request an alternate arrangement and due date. If an alternate arrangement is not requested or offered then no credit will be given for the assignment. Any approved make-up assignments should be submitted to the instructor through Canvas mail.

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 me within 24 hours of the technical difficulty if you wish to request a make-up.

Policy Related to Required Class Attendance

This is an online course and therefore, attendance means you are expected to complete the course learning activities so that you meet the established deadlines. 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>

Students who wish to drop from the course must do so by the drop/add deadline established by the Office of the University Registrar. Students must not assume they will be automatically dropped if they fail to participate in the course learning activities.

STUDENT EXPECTATIONS, ROLES, AND OPPORTUNITIES FOR INPUT

Communication Guidelines

All faculty, staff, and students of the University are required and expected to obey the laws and legal agreements governing software use. Failure to do so can lead to monetary damages and/or criminal penalties for the individual violator. Because such violations are also against University policies and rules, disciplinary action will be taken as appropriate. We, the members of the University of Florida community, pledge to uphold ourselves and our peers to the highest standards of honesty and integrity.

Message Board Netiquette and Guidelines

- Treat instructor with respect, even via Canvas or in any other online communication
- Always use your professors' proper title: Dr. or Prof.
- Use clear and concise language
- Remember that all communication should have correct spelling and grammar
- Use standard fonts such as Times New Roman and use a size 12 or 14 pt. font
- Avoid the use of emoticons like :)
- Be cautious when using humor or sarcasm as tone is sometimes lost in a Canvas discussion post and your message might be taken seriously or offensive
- Be careful with personal information (both yours and other's)
- Do not send confidential information via e-mail

For Posting on-line:

- Make posts that are on topic and within the scope of the course material
- Review and edit your posts before sending
- Be as brief as possible
- Provide citations if appropriate
- Be sure to read all messages in a thread before replying
- Avoid short, generic replies such as, "I agree." You should include why you agree or add to the previous point
- Express your opinions in a respectful, non-critical way
- Be open-minded

Student Privacy

There are federal laws protecting your privacy with regards to grades earned in courses and on individual assignments. For more information, please see:

<http://registrar.ufl.edu/catalog0910/policies/regulationferpa.html>

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.

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, 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.

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

Campus Resources:

Academic Resources

E-learning technical support, 352-392-4357 (select option 2) or e-mail to Learning-support@ufl.edu.

<https://lss.at.ufl.edu/help.shtml>.

Career Resource Center, Reitz Union, 392-1601. Career assistance and counseling.

<https://www.crc.ufl.edu/>.

Library Support, <http://cms.uflib.ufl.edu/ask>. Various ways to receive assistance with respect to using the libraries or finding resources.

Teaching Center, Broward Hall, 392-2010 or 392-6420. General study skills and tutoring.

<https://teachingcenter.ufl.edu/>.

Writing Studio, 302 Tigert Hall, 846-1138. Help brainstorming, formatting, and writing papers.

<https://writing.ufl.edu/writing-studio/>.

Student Complaints Campus: https://www.dso.ufl.edu/documents/UF_Complaints_policy.pdf.

On-Line Students Complaints: <http://www.distance.ufl.edu/student-complaint-process>.