**EPI 690G: Genetic Epidemiology**

**Spring 2021**

*(version 2.0)*

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| **Contact Me**  https://static.thenounproject.com/png/551101-200.png | **Meet with Me**  https://static.thenounproject.com/png/947025-200.png | **Location and Time**  https://static.thenounproject.com/png/2511569-200.png | **Course Materials**  https://static.thenounproject.com/png/349329-200.png |
| Cassandra Spracklen, PhD  Pronouns: she, her, hers  [cspracklen@umass.edu](mailto:cspracklen@umass.edu) (Please allow 24 hours for response, not including weekends) | Please email me for an appointment | Remote via Zoom  <https://umass-amherst.zoom.us/j/92098416005>  Tuesday & Thursday 11:30am-12:45pm | No required textbook  All required reading and other course materials will be posted on our Moodle page. |

**General Course Description:**

Genetic Epidemiology is a field of study that deals with the genetic etiology and distribution of diseases in populations. This course will provide students with a focused exposure to genetic analysis, with a major emphasis on association analysis. Topics will include different approaches to measuring the association of genes with disease: family history, heritability, and genetic association, how to model gene-by-environment interactions, epigenetics, and Mendelian Randomization as an approach to causal inference. Students will be exposed to the tools needed to critically review the literature in genetic epidemiology and human genetics. Issues related to study implementation are considered. We will also cover some of the ethical, social, and legal implications of using genetics in epidemiology and public health.

**Prerequisites:**

EPI 630 or instructor approval

**Course objectives:**

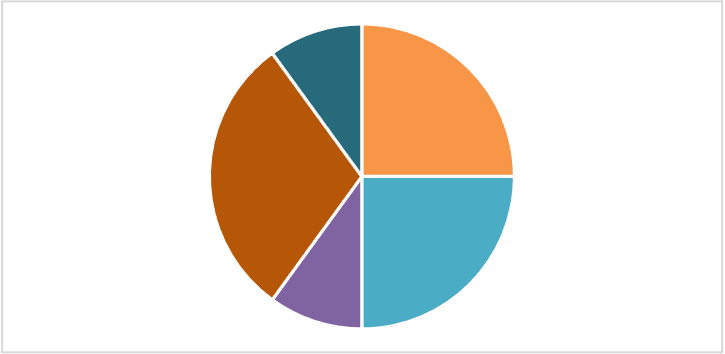
After completing this course, you will:

* Be able to properly use and understand the vocabulary associated with genetic epidemiology
* Have an understanding of the goals, analytical methods, and challenges of genetic epidemiology
* Know the methods that are relevant to studies of genes in individuals, populations, and families, including methods used in population genetics, quantitative trait genetics, studies of familial aggregation and heritability, association studies, gene x gene and gene x environment interaction studies, and linkage analysis
* Be familiar with a variety of sampling designs, data collection methods, and practical difficulties, including statistical power, associated with fielding a genetic epidemiology study
* Have an expanded perspective on disease etiology
* Understand the ethical, social, and legal implications of using genetics in epidemiology and public health

**Course Format:**

Our class meetings will involve a combination of lectures, readings, in-class activities, and group discussions. In class, we will apply the concepts covered in the reading assignment during that day. Therefore, completing the reading on time is **essential** for success in the course.

**Course Requirements and Grading:**



Hot Topic Assignments

Genetics Research Paper

In-Class Activities

Health Topic & Problem Statement

Presentation

*Hot Topic Assignments (25% of course grade):* As a field, Genetic Epidemiology research is full of ethical, social, and/or legal implications (aka, “hot topics”). For Hot Topic assignments, you will complete assigned readings and address specific questions prior to coming to class. The specific questions may: be a summary of current stakeholder sides of the argument, ask you to write your own opinions on the topic, or may be related to your selected health topic area of interest (*see Health Topic assignment below*). Assigned readings and specific questions to address for each Hot Topic will be posted to Moodle at least one week before it is due. Specific instructions for each Hot Topic assignment may vary and will be provided on each assignment description sheet. Hot Topic assignments should be submitted electronically as Microsoft Word Documents or PDFs to Moodle. Email assignments to the instructor will not be accepted.

Hot Topic Assignments will, in general, have an accompanying class discussion (as noted Moodle). You must complete the hot topic assignment, turn in to Moodle, AND attend the class of any subsequent discussion to full earn points for the assignment. Hot Topic assignments and participation in the following discussion will generally be worth 10 points. The grading rubric for the Hot Topic Readings can be found with the assignment.

*In-Class Assignments (25% of course grade):* We will have various in-class worksheets and activities throughout the semester. Dates of these activities will be posted on Moodle and the course syllabus. You must attend the in-class activity and participate to earn points. 5 points will be assigned for participation in each in-class activity. In the case of an excused absence, you may make-up these points by requesting a make-up assignment as soon as possible and no later than 1 week after the missed class period. Points may be deducted from the daily total if you do not meet expectations as described in the following rubric:

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| --- | --- | --- | --- | --- |
| **Expectations** | **Met expectations** | **Points** | **Did not meet expectations** | **Points** |
| Class conduct and respect for others | Respectful and considerate to others | 1 | * Disruptive during class * Distracts others * Engages in unrelated activities | 0 |
| Class involvement  (each bullet is worth 1 point). | * Makes appropriate contributions to In-class activity * Alert and on task * Uses time productively when working in pairs or groups * Participates fully as a member of a group | 4 | * Comes to class unprepared * Seldom contributes or listens to others * Is distracted and not using time appropriately * Makes little or no contribution in pairs or groups | 0 |

*Health Topic Problem Statement (10% of grade):* You will prepare a 2-3 page summary of your selected health topic as a precursor for your final genetics research paper. This assignment provides you with an opportunity to get instructor feedback on the content you anticipate including in your final Genetics Research Paper. Assignment description, formatting details, suggested content, and grading rubric for the Genetics Research Paper can be found on Moodle.

*Genetics Research Paper (30% of grade):* You will prepare a genetics research paper focused on your assigned health topic. This paper will be a culmination of what you have learned during this semester during the lectures, in-class assignments, and hot topic assignments, and it should be an extension of your health topic and problem statement. You should be sure to include a short literature review of the topic. Assignment description, formatting details, suggested content, and grading rubric for the Genetics Research Paper can be found on Moodle.

Many of you will be writing and/or presenting MS theses or MPH projects during the latter half of this semester (with varying deadlines). Others will be preparing for their PhD comprehensive exams in May. To help you plan for your semester (and to try and limit your stress!), this assignment has a flexible due date between April 16-May 11. This means that I will accept your papers at any time during this window. Please plan accordingly based on your schedule.

*Student Presentations (10% of grade):* You will give 10-minute PowerPoint presentations of your genetics research paper. Specific guidelines for the presentation will be posted on Moodle. Presentation dates for each student will be determined in late March or early April.

**Final Course Grade**: The relationship between your course percentage and final letter grades is as follows:

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| --- | --- | --- | --- | --- | --- | --- | --- |
| Letter Grade | Percent | | Letter Grade | Percent | Letter Grade | Percent | |
| A | ≥ 93% | B- | | 80-82.9 | D+ | 67-69.9 |
| A- | 90-92.9 | C+ | | 77-79.9 | D | 63-66.9 |
| B+ | 87-89.9 | C | | 73-76.9 | F | <63 |
| B | 83-86.9 | C- | | 70-72.9 |  |  |

**REQUIRED TEXTS AND TOOLS:**

**Textbook:**

No required textbook.

Recommended (particularly for those who may be doing any genetic data analyses): *An Introduction to Statistical Genetic Data Analysis* (ISBN: 9780262538381) Melinda Mills, Nicola Barban, and Felix Tropf: 2020.

**Lecture notes:**

Lecture notes will be posted to the course website prior to each class period. A course schedule appears at the end of this syllabus.

**Other instructional materials:**

All additional course materials will be posted to the course website or distributed in class as needed.

**Information to Support your Success in this Course:**

*Attendance:* Arriving on time to class will help ensure its success and also shows respect to your classmates. It is important that you attend every class because we engage in many learning activities during class time. It’s also hard to participate in the discussion if you aren’t present for it.

*Laptops, Cell Phones and other Electronics:*Please be courteous to your fellow students and refrain from using your cell phones or other personal electronic devices during class time. You are welcome to use your laptops or tablets. You are asked to refrain from accessing email, social media, or non-course related websites during class. Avoiding these distractions will help you fully participate in the classroom discussions.

*Website:*I will post all course materials on Moodle, and you will submit all assignments via Moodle or our class Google Drive. Because Moodle and Google Drive are important class resources, make sure you have access to it early in the semester. If you have trouble accessing the course website, please let me know as soon as possible.

*Late Assignments:* Assignments submitted after 11:30am on the due date will be considered late. 10% of the points will be deducted for each day the assignment is late. Contact Dr. Spracklen in advance if you have an *exceptional circumstance* (for which appropriate documentation is required) as to why you were unable to submit your assignment on time.

*Extra Credit:* Although unlikely, I may provide opportunities for students to earn extra credit during the semester; if so, details will be posted on Moodle and discussed in class.

*Student Hours:*I am always happy to answer brief questions immediately before or after class. Please email me if you would like to meet at a different time.

*Accommodation and Inclusive Learning Statement:*If you have a disability and require accommodations, please let me know as soon as possible. You will need to register with Disability Services (161 Whitmore Administration Building; phone (413) 545-0892). Information on services and materials for registering are also available on their website: [www.umass.edu/disability](http://www.umass.edu/disability) .

Your success in this class is important to me. We all learn differently and bring different strengths and needs to the class. If there are aspects of the course that prevent you from learning or make you feel excluded, please let me know as soon as possible. Together we’ll develop strategies to meet both your needs and the requirements of the course. There are also a range of resources on campus, including:

* Writing Center: [www.umass.edu/writingcenter](http://www.umass.edu/writingcenter)
* Learning Resource Center: [www.umass.edu/lrc](http://www.umass.edu/lrc)
* Center for Counseling and Psychological Health: [www.umass.edu/counseling](http://www.umass.edu/counseling)
* English as a Second Language Program: [www.umass.edu/esl](http://www.umass.edu/esl)

*Academic Honesty:* We want our learning environment to be honest and fair. UMass Amherst has an Academic Honesty Policy that includes cheating and plagiarism as forms of dishonesty, among others. You can read the full policy and find other helpful resources here: <https://www.umass.edu/honesty/resources> .

If you are unsure as to what actions specifically violate the academic honesty code, contact me immediately for clarification.

*Email communications*: I will respond to emails within 24 hours of receiving them from you. I will respond to emails sent between Friday 4pm-Sunday evening on Monday.

*Valuing, Recognizing, and Encouraging Diversity***:** Promoting and valuing diversity in the classroom enriches learning and broadens everyone’s perspectives. Inclusion and tolerance can lead to respect for others and their opinions and is critical to maximizing the learning that we expect in this course. Our own closely held ideas and personal comfort zones may be challenged. The results, however, create a sense of community and promote excellence in the learning environment. Diversity includes consideration of (1) the variety of life experiences others have had, and (2) factors related to “diversity of presence,” including age, economic circumstances, ethnic identification, disability, gender, geographic origin, race, religion, sexual orientation, social position. This class will follow principles of inclusion, respect, tolerance, and acceptance that support the values of diversity.

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| **COURSE CALENDAR** |

*These dates are subject to change at the discretion of the instructor.*

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| **Day** | **Date** | **Agenda/Topic** | **Reading(s)/Video(s)** | **Assignment(s) Due** |
| *I. Genetic Concepts and Genetic Basis for Disease* | | | | |
| Tuesday | 2/2 | Class Introduction |  |  |
| Thursday | 2/4 | Overview of Genetic Epidemiology | Duggal *AJE* 2019 |  |
| Tuesday | 2/9 | Chromosomal Basis of Disease | AS: Chromosomes |  |
| Thursday | 2/11 | Prenatal Testing | Richardson *SFNM* 2018 Johnson *NEJM* 2017 | Hot Topic #1 |
| Tuesday | 2/16 | Single Gene Inheritance | AS: Alleles and Genes; Monohybrids; Pedigree |  |
| Thursday | 2/18 | DNA Mutations and Repair | AS: Protein Synthesis; Mutations |  |
| Tuesday | 2/23 | Gene Therapy and CRISPR | You *MTMCD* 2019 Cyranoski *Nature* 2019 Kofler *Nature* 2019 | Hot Topic #2 |
| Thursday | 2/25 | Population Genetics: HWE | *AS: HWE* |  |
| Tuesday | 3/2 | Population Genetics: Admixture and LD | *(2) YouTube videos posted to Moodle* |  |
| Thursday | 3/4 | Race and Genetics | Caulfield *GM* 2009  Borrell *NEJM* 2021 | Hot Topic #3 |
| *II. Genetic Associations and Applications* | | | | |
| Tuesday | 3/9 | Genetics of Complex Traits and Disease |  |  |
| Thursday | 3/11 | Performing a GWAS (Genotyping, Data Cleaning, Imputation, Interpreting Results) | *Intro to Statistical Genetic Data Analysis* Chapter 4 |  |
| Tuesday | 3/16 | Post GWAS: Meta-Analyses, Multi-ancestry Studies, Functional Annotation | Visscher *AJHG* 2017  Loos *Nat Comm* 2020 |  |
| Thursday | 3/18 | GWAS Journal Club | Voight *Nat Genet* 2012 | JC Worksheet |
| Tuesday | 3/23 | Extensions and Applications of GWAS: GxE / MR / Pleiotropy / PheWAS | Davies *BMJ* 2018 |  |
| Thursday | 3/25 | Annotating a GWAS Variant |  |  |
| Tuesday | 3/30 | Era of OMICS | Hasin *Genome Biol* 2017 |  |
| *III. Genetic Epidemiology in Public Health / Genetic Medicine* | | | | |
| Thursday | 4/1 | Direct-to-Consumer Testing and Forensic Geneticists | Oh *Genom Inform* 2019 |  |
| Sunday | 4/4 | **Health Topic Problem Statement Due by 11:59pm** | | |
| Tuesday | 4/6 | *No Class – “Work Day”!!* | | |
| Thursday | 4/8 | Polygenic Risk Scores Journal Club | Said JAMA 2018 | JC Worksheet |
| Tuesday | 4/13 | Forensics (Guest Lecture) |  |  |
| Thursday | 4/15 | Genetic Privacy | Weaver *Bioethics* 2016 HD Fact Sheet | Hot Topic #4 |
| Tuesday | 4/20 | *No class—Wednesday Schedule* | | |
| Thursday | 4/22 | Presentations |  |  |
| Tuesday | 4/27 | Presentations |  |  |
| Thursday | 4/29 | Presentations |  |  |
| Tuesday | 5/4 | Presentations |  |  |
| Tuesday | 5/11 | Finals Week – No Class  **Genetics Research Paper Due by 11:59pm** (Flexible Due Date 4/16-5/11) | | |

*AS: Ameoba Sisters (YouTube Links on Moodle)*