

Mel and Enid Zuckerman College of Public Health

University of Arizona

**SYLLABUS**

EPID 677 - Principles of Genetic Association Studies

# Spring 2019

**Time**: Monday, 9:00 – 11:50 am

**Location**: Drachman Hall, A119 & MEZCOPH Computer Lab, Drachman Rm A319

**Instructor**: Yann Klimentidis, PhD

MRB, rm 115

Phone: (520) 621-0147

Email: yann@email.arizona.edu

**Office Hours**: by appointment – please email or call me.

**Teaching Assistant**: None

**Course Description:**

The course will focus on the principles, methods, and challenges involved in dissecting the genetics of complex diseases using association studies. It will consist of a theoretical component and a hands-on, applied component (using R software for genetic data analysis in a computer lab). Specific topics will be: Heritability; Population genetics, Linkage disequilibrium and population stratification; Epidemiological design strategies for genetic association studies; GWAS, sequencing, gene-by-environment interactions, genetic risk prediction, epigenetics, mendelian randomization, and use of online genetic databases.

**Course Prerequisites:**

EPI573A and EPI576A, or some background in epidemiology and/or statistics are strongly suggested, but not required.

**Course Learning Objectives:**

To provide an overview of the concepts, methods, and hands-on applications to design, conduct, and interpret genetic association studies and to conduct genotype analyses.

**MPH/Program Competencies Covered:**

Core MPH competencies that will be addressed during this course in include: A1-7; B1-2; D1,3,4,6; E 1,3

These are available on the College’s Website at: <https://publichealth.arizona.edu/sites/publichealth.arizona.edu/files/academics/MPH%20Competencies.pdf>

**Course Notes:**

Lecture presentations and reading material will be posted on D2L.

**Class Attendance/Participation**: Students are expected to attend lectures and participate actively in discussion and in group activities during class.

**Course Requirements:**

Class attendance; read assigned paper and complete paper dissection questions prior to class; co-lead paper presentation/discussion; take quizzes on lecture material; completion of in-class R exercises

**Quizzes:** We will have a quiz on most days that we are in A119 (i.e. not computer lab week). The quiz will cover the material from the previous lecture. You will take the quiz on D2L on your laptop or phone. I will allow you to drop your lowest grade.

**Article dissections:** These will be the same for each paper that we discuss on lecture weeks. They will be due BEFORE class. You will be asked to identify the hypotheses and predictions, describe the methods, results, and conclusions, and provide your assessment of the paper/study. I expect about a 2-3 sentence response per question. Your grade will be based on answering the question correctly, original and creative thought on your part, level of critical evaluation and evidence of reading the paper thoroughly. I will allow you to drop your lowest grade.

**Group presentation & leading discussion:** The first week of class you will sign up for a week to lead a paper discussion. Your role for these discussions is not to talk the whole time, but to provide a little bit of background and then pose thoughtful questions that can be discussed in class. Each member of the group should present/lead discussion an equal amount.

**Groups should email the instructor indicating the paper that they have chosen on the Monday of the week prior to your presentation.**

For background: do not summarize the article or go into much depth about what was in the article (everyone should have read it). Instead try to put the article into a greater context. What do we already know about the general topic? Why is it interesting? Briefly describe what other studies have found (a good place to find references to other studies is in the introduction where the background is discussed-look up these papers and read the abstract, look at the figures, maybe skim through the methods).

Discussion: try to start with the basics: ask the class what they thought the main hypotheses and conclusions were. Ask them if they agree with the conclusions and what they thought about the methods, etc. Then try to have 3-4 additional questions designed to promote discussion in class. Debates are a good idea. You should also copy and paste figures from the results section onto slides for discussion.

Power point: make a short power point presentation to help you lead discussion. It should include:

-2-5 slides on background

-a slide for each table and figure (just copy and paste these in)

-a slide for each of your discussion questions

Presentation suggestions:

• First provide a brief summary of background information that is necessary to understand the paper.

• Potential questions to pose when leading discussions:

• Why did the author’s conduct this study?

* How does this study contribute to our knowledge of genetic epidemiology ?

• How was the study relevant to what we have covered in class?

• What were the author’s hypotheses? Predictions?

• Why did the authors choose this particular population?

• What are the main findings? (here it might be a good idea to go present figures).

• How do these findings relate to the hypotheses/research topic?

• What aspects of the study did the authors deal with well? Why do you think that was a good way to handle the research topic?

• Are any parts of the study controversial? Explain.

• How could the study be improved?

• What are some future studies that could be done along these lines/on this topic?

**R exercises**: You are expected to complete R-based exercises in which you will analyze genetic and phenotypic data. These in-class R exercises must be turned in at the end of class, unless otherwise notified.The lowest in-class exercise will be dropped.

**Grading:**

in-class R exercises: 25%; Discussion lead: 15%; Quizzes: 25%; Participation/Group activities: 15%; paper dissection: 20%.

Grading scale: 100-90: A; 80-89: B; 70-79: C; 60-69: D; below 60: F. This scale may be revised by instructor, if necessary.

**Communications**: You are responsible for reading emails sent to your UA account from your instructor and the announcements that are placed on the course web site. Information about readings, news events, your grades, assignments and other course related topics will be communicated to you with these electronic methods. The official policy can be found at: <https://www.registrar.arizona.edu/personal-information/official-student-email-policy-use-email-official-correspondence-students>

**Disability Accommodations:**

**It is the University’s goal that learning experiences be as accessible as possible.  If you anticipate or experience physical or academic barriers based on disability or pregnancy, please let me know immediately, so that we can discuss options. You are also welcome to contact the Disability Resources (520-621-3268) to establish reasonable accommodations (as it is very important that you be registered with the DRC). For additional information on Disability Resources and reasonable accommodations, please visit** <http://drc.arizona.edu/students>

**Code of Academic Integrity**

Students are encouraged to share intellectual views and discuss freely the principles and applications of course materials. However, graded work/exercise must be the product of independent effort unless otherwise instructed. Students are expected to adhere to the UA Code of Academic Integrity, available through the office of the UA Dean Students: <http://deanofstudents.arizona.edu/policies-and-codes/code-academic-integrity>

**Classroom Behavior**: (Statement of expected behavior and respectful exchange of ideas:

Present policies to foster a positive learning environment, including use of cell phones, mobile devices, etc.).

Students are expected to be familiar with the UA Policy on Disruptive Student Behavior in an Instructional Settingfound at: <http://policy.arizona.edu/education-and-student-affairs/disruptive-behavior-instructional-setting>

**Threatening Behavior Policy**: The UA Threatening Behavior by Students Policy prohibits threats of physical harm to any member of the University community, including to one’s self, <http://policy.arizona.edu/education-and-student-affairs/threatening-behavior-students>

**Nondiscrimination and Anti-Harassment Policy:**

The University of Arizona is committed to creating and maintaining an environment free of discrimination, <http://policy.arizona.edu/human-resources/nondiscrimination-and-anti-harassment-policy>

**UA Smoking and Tobacco Policy:**

The purpose of this Policy is to establish the University of Arizona’s (University) commitment to protect the health of University faculty, staff, students, and visitors on its campuses and in its vehicles,  
<http://policy.arizona.edu/ethics-and-conduct/smoking-and-tobacco-policy>

**Syllabus Changes:**  Information contained in the course syllabus, other than the grade and absence policies, may be subject to change with reasonable advance notice, as deemed appropriate by the instructor.

**COURSE SCHEDULE** (may be revised):

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| --- | --- | --- | --- | --- | --- | --- |
|  | Date | Location | Topic | Assignment due at the end of class | Quiz | Discussion paper |
| 1 | January 14 | Computer Lab - A319 | Syllabus, introduction, introduction to R and genetic data |  |  |  |
| **2** | January 28 | A119 | Basic molecular genetics / Heritability |  |  | TBD |
| **3** | February 4 | Computer Lab - A319 | Basic genetic dataset analysis in R | Exercise 1 |  |  |
| **4** | February 11 | A119 | Population genetics / Linkage disequilibrium |  | Quiz #1 | TBD |
| **5** | February 18 | Computer Lab - A319 | Online genetic databases; data quality control; population stratification | Exercise 2 |  |  |
| **6** | February 25 | A119 | Genetic association studies I |  | Quiz #2 | TBD |
| **7** | March 11 | Computer Lab - A319 | Statistical tests for genetic association | Exercise 3 |  |  |
| **8** | March 18 | A119 | Genetic association studies II |  | Quiz #3 | TBD |
| **9** | March 25 | Computer Lab - A319 | Statistical tests for genetic association, cont. | Exercise 4 |  |  |
| **10** | April 1 | A119 | Gene-by-Environment interactions |  | Quiz #4 | TBD |
| **11** | April 8 | Computer Lab - A319 | Genetic risk scores, gene-by-environment interactions | Exercise 5 |  |  |
| **12** | April 15 | A119 | Missing heritability & prediction |  | Quiz #5 | TBD |
| **13** | April 22 | Computer Lab - A319 | Genomic prediction analysis: basic concepts and applications | Exercise 6 |  |  |
| **14** | April 29 | A119 | Mendelian randomization, Epigenetics, future trends |  | Quiz #6 | TBD |