Mel and Enid Zuckerman College of Public Health
University of Arizona

SYLLABUS
EPID 677 - Principles of Genetic Association Studies
Spring 2016

Time: Thursday, 4:00 – 6:50 pm

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

Teaching Assistant: None

Course Description:
The course will focus on the principles, methods, and challenges to dissect 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; Linkage disequilibrium and population stratification; Epidemiological design strategies for genetic association studies; Allele, genotype, and haplotype analyses; GWAS, sequencing, and integration with other biomarkers; Gene-by-environment interactions, genomic prediction, epigenetics, mendelian randomization, and use of online genetic databases.

Course Prerequisites:
EPI573A and EPI576A, or approval by instructors.

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.

Course Notes:
Hand-outs and papers of interest will be distributed in class and/or posted electronically.

Discussion Papers:
We will discuss assigned papers every other week as indicated in the syllabus, in the second half of the class period. Students are expected to read the paper before class and
participate in class discussion. Specific papers are subject to change during the semester in which case students will be notified.


**Recommended (not required) Text:**

**Alternative Texts:**
- Duncan Thomas: “Statistical Methods in Genetic Epidemiology”, Oxford University Press

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

**Course Requirements:**
Class attendance; read and discuss Discussion papers; completion of in-class exercises; Mid-term in-class exam; one paper critique presentation (guidelines will be distributed in class); and a final exam.

**Grading:**
Midterm exam: 20%; in-class exercises: 25%; final exam: 20%; paper critique: 15%; participation: 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. The lowest in-class exercise will be dropped. In-class exercises must be turned in at the end of class, unless otherwise notified.

Spring 2016
Communications: You are responsible for reading emails sent to your UA account from your professor 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: http://www.registrar.arizona.edu/emailpolicy.htm

Disability Accommodation: If you anticipate issues related to the format or requirements of this course, please meet with me. I would like us to discuss ways to ensure your full participation in the course. If you determine that formal, disability-related accommodations are necessary, it is very important that you be registered with Disability Resources (621-3268; drc.arizona.edu) and notify me of your eligibility for reasonable accommodations. We can then plan how best to coordinate your accommodations. The official policy can be found at: http://catalog.arizona.edu/2012%2D13/policies/disability.htm

Academic Integrity: All UA students are responsible for upholding the University of Arizona Code of Academic Integrity, available through the office of the Dean of Students and online: The official policy found at: http://deanofstudents.arizona.edu/codeofacademicintegrity

Classroom Behavior: (Statement of expected behavior and respectful exchange of ideas) The Dean of Students has set up expected standards for student behaviors and has defined and identified what is disruptive and threatening behavior. This information is available at: http://deanofstudents.arizona.edu/disruptiveandthreateningstudentguidelines

Grievance Policy: Should a student feel he or she has been treated unfairly, there are a number of resources available. With few exceptions, students should first attempt to resolve difficulties informally by bringing those concerns directly to the person responsible for the action, or with the student's graduate advisor, Assistant Dean for Student and Alumni Affairs, department head, or the immediate supervisor of the person responsible for the action. If the problem cannot be resolved informally, the student may file a formal grievance using the Graduate College Grievance Policy found at: http://grad.arizona.edu/academics/policies/academic-policies/grievance-policy

Grade Appeal Policy: http://catalog.arizona.edu/2012-13/policies/gradappeal.htm

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

<table>
<thead>
<tr>
<th>Date</th>
<th>Location</th>
<th>Topic</th>
<th>Assignment due at the end of class</th>
<th>Discussion Paper</th>
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<tbody>
<tr>
<td>1 January 14</td>
<td>Computer Lab - A319</td>
<td>Syllabus, introduction, introduction to R and genetic data</td>
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<tr>
<td>2 January 21</td>
<td>A119</td>
<td>Basic molecular genetics / Heritability</td>
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<tr>
<td>3 January 28</td>
<td>Computer Lab - A319</td>
<td>Basic genetic dataset analysis in R</td>
<td>Exercise 1</td>
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<td>4 February 4</td>
<td>A119</td>
<td>Population genetics / Linkage disequilibrium</td>
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<tr>
<td>5 February 11</td>
<td>Computer Lab - A319</td>
<td>Online genetic databases; data quality control; population stratification</td>
<td>Exercise 2</td>
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<td>6 February 18</td>
<td>A119</td>
<td>Genetic association studies I</td>
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<td>7 February 25</td>
<td>Computer Lab - A319</td>
<td>Statistical tests for genetic association</td>
<td>Exercise 3</td>
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<td>8 March 3</td>
<td>A119</td>
<td>In-class midterm examination / genetic association studies II</td>
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<td>9 March 10</td>
<td>A119</td>
<td>Gene-by-Environment interactions</td>
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<td>10 March 24</td>
<td>Computer Lab - A319</td>
<td>Statistical tests for genetic association, cont.</td>
<td>Exercise 4</td>
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<td>11 March 31</td>
<td>A119</td>
<td>Missing heritability, prediction, sequencing data</td>
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<td>12 April 7</td>
<td>Computer Lab - A319</td>
<td>Genetic risk scores, gene-by-environment interactions</td>
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<td>13 April 14</td>
<td>A119</td>
<td>Mendelian randomization, Epigenetics, future trends</td>
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<tr>
<td>14 April 21</td>
<td>Computer Lab - A319</td>
<td>Genomic prediction analysis: basic concepts and applications</td>
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<td>15 April 28</td>
<td>A119</td>
<td>Paper critique presentations</td>
<td>Copy of slide presentation</td>
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<td>May 11</td>
<td>A119</td>
<td><strong>Final Exam 6:00 – 8:00 PM</strong></td>
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