Mel and Enid Zuckerman College of Public Health
University of Arizona

SYLLABUS
Principles of Genetic Association Studies
[Prin Genetic Assoc Stds – EPID677]

Summer II 2010

Time: MoTuWeThFr, 1pm - 3.50pm

Location: Drachman Hall, A122

Instructor: Stefano Guerra, MD, PhD
AHSC, 2348A1
Phone: (520) 626-7411;
Email: sguerra@arc.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. Specific topics will be: Epidemiological design strategies for genetic association studies; Selection of positional and/or functional candidate genes; Linkage disequilibrium and population stratification; Allele, genotype, diplotype, and haplotype analyses; GWAS and Mendelian randomization; Gene-by-environment and gene-gene interactions.

Course Prerequisites:
EPI573A, EPI576A, and EPI576B

Course Learning Objectives:
To provide an overview of the concepts and methods to design and interpret genetic association studies and to conduct genotype and haplotype analyses.

EPI Competencies:

Domain 1: General Knowledge
Describes risk factors for well established health problems and the evidence in support of these factors
Understands basic research designs used in health issues
Domain 2: Problem Identification / Planning
Defines a research or health problem by identifying gaps in the knowledge of a health issue
Identifies steps needed to accurately assess and understand the health status of populations
Identifies determinants of health and illness, factors contributing to health promotion and disease prevention, and factors influencing the use of health services
Domain 3: Information Skills
Retrieves and organizes literature from valid sources of the evidence base
Judges, critiques, and interprets research findings
Domain 4: Communication
Organizes and delivers oral presentations of research findings or health issues in varying professional formats
Organizes and writes a manuscript suitable for publication in peer-reviewed publications
Domain 6: Professional Conduct
Recognizes and responds appropriately to ethical issues in the conduct of scientific research
Domain 8: Study Design
Selects and defines measures and variables relevant to defined health problems
Domain 9: Study Conduct
Develops strategies for monitoring study progress and preparing status reports
Identifies strategies for recruitment and retention of study participants and how these strategies can be adapted to account for cultural differences
Domain 11: Data Analysis
Determines appropriate uses and limitations of data
Domain 12: Data Interpretation
Evaluates the integrity, comparability of data, and limitations of data
Makes relevant inferences from data analyses
Understands and applies the principles of causality
Identifies areas of further research

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

Recommended Text:

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

Course Requirements:
Mid-term in-class exam; homeworks; one paper critique.

Grading:
Homework 15%; paper critique 35%; midterm: 35%; participation: 15%.

Class Attendance/Participation: Students are expected to attend lectures and participate actively to discussion.
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/2008%2D09/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/policiesandcodes/codeofacademicintegrity

Classroom Behavior: (Statement of expected behavior and respectful exchange of ideas) Students are expected to be familiar with the UA Policy on Disruptive Behavior in an Instructional Setting found at: http://web.arizona.edu/~policy/distuptive.pdf

and the Policy on Threatening Behavior by Students found at: http://web.arizona.edu/~policy/threatening.pdf

Grade Appeal Policy: http://catalog.arizona.edu/2009-10/policies/gradappeal.htm

Graduate Student Grievance Policy: http://grad.arizona.edu/academics/policies/academic-policies/grievance-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.
# Course Schedule (may be revised):

<table>
<thead>
<tr>
<th>Date / location</th>
<th>Topic</th>
<th>Lecturer</th>
<th>Assignment due</th>
<th>Books / papers</th>
</tr>
</thead>
<tbody>
<tr>
<td>1 July 12 / A122</td>
<td>Syllabus, introduction, the nuts and bolts of genetic association studies</td>
<td>S Guerra</td>
<td>Notes (class 1) / Paper 1</td>
<td>ZG Chpt 1 / Notes (class 2)</td>
</tr>
<tr>
<td>2 July 13 / A122</td>
<td>Basic molecular genetics</td>
<td>P Thompson</td>
<td>ZG Chpt 1 / Notes (class 2)</td>
<td>any epi book / Notes (class 3)</td>
</tr>
<tr>
<td>3 July 14 / A122</td>
<td>Epidemiological principles</td>
<td>S Guerra</td>
<td>ZG Chpt 4 / Notes (class 4)</td>
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<tr>
<td>4 July 15 / A122</td>
<td>Population genetics</td>
<td>FJ Meaney</td>
<td>ZG Chpt 4 / Notes (class 4)</td>
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</tr>
<tr>
<td>5 July 16 / A122</td>
<td>Hmw 1 review / terminology / Mendelian inheritance / linkage disequilibrium (LD)</td>
<td>S Guerra</td>
<td>Hmw 1</td>
<td>ZG Chpt 2, 3, 9 / Notes (class 5)</td>
</tr>
<tr>
<td>6 July 19 / A122</td>
<td>Hmw 2 review / Hardy-Weinberg Equilibrium / LD II / genetic association studies</td>
<td>S Guerra</td>
<td>Hmw 2</td>
<td>ZG Chpt 3, 9, 10 / Notes (class 6)</td>
</tr>
<tr>
<td>7 July 20 / A122</td>
<td>In-class midterm examination / genetic association studies II</td>
<td>S Guerra</td>
<td>ZG Chpt 10, 12 / Notes (class 7)</td>
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<tr>
<td>8 July 21 / A122</td>
<td>Midterm review / genetic association studies III / new approaches</td>
<td>S Guerra</td>
<td>ZG Chpt 10, 12 / Notes (class 8)</td>
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<tr>
<td>9 July 22 / A122</td>
<td>Homework 3 review / Phenotypic characterization / Gene-by-Environ.: principles / GWAS: principles</td>
<td>S Guerra / D Stern</td>
<td>Hmw 3</td>
<td>Notes (class 9)</td>
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<tr>
<td>10 July 23 / A122</td>
<td>Gene-by-Environment: applications / GWAS: applications</td>
<td>S Guerra / C Rebordosa</td>
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<td>Notes (class 10)</td>
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<tr>
<td>11 July 26 / Drachman B206</td>
<td>Online Genetic Databases</td>
<td>W Klimecki</td>
<td>Notes (class 11)</td>
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<tr>
<td>12 July 27 / computer lab</td>
<td>Hmw 4 review / Phase resolution / statistical power computation</td>
<td>S Guerra</td>
<td>Hmw 4</td>
<td>Phase / Quanto documentation</td>
</tr>
<tr>
<td>13 July 28 / A122</td>
<td>GxG pathway analysis / Mendelian Randomization / Hmw 5 review</td>
<td>LeRoy / Guerra</td>
<td>Hmw 5</td>
<td>Notes (class 13)</td>
</tr>
<tr>
<td>14 July 29 / A122</td>
<td>Biomarkers / overview</td>
<td>S Guerra / Forum</td>
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<td>Notes (class 14)</td>
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<tr>
<td>15 July 30 / A122</td>
<td>Paper critique presentations</td>
<td>Students</td>
<td>Written paper critique due</td>
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