PUBHLTH 690T: Applied Statistical Genetics
Spring 2015, MoWe 4:00PM-5:15PM

Instructor:
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Office Hours: Tu 10:00AM-12:00PM

Teaching Assistant:
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Requirements:
- Elementary knowledge of statistical methods at the level of a first course in biostatistics.
- Familiarity with the statistical software package R (http://cran.r-project.org/).

Course Goals: The aim of this course is to provide students with fundamental statistical concepts and tools relevant to the analysis of high-dimensional genomics data arising from population-based association studies. Students will learn to program and implement the techniques using R.

Required Textbook:
Applied Statistical Genetics with R: For Population-based Association Studies
by Andrea S. Foulkes, Springer (2009); ISBN: 978-0-387-89553-6
eBook:
http://silk.library.umass.edu/login?url=http://dx.doi.org/10.1007/978-0-387-89554-3

Recommended supplemental material:
- Fundamentals of Biostatistics by B. Rosner, Cengage Learning (2010).
- A Statistical Approach to Genetic Epidemiology by A. Ziegler and I. Konig, Wiley-VCH (2007).
- An Introduction to R: http://cran.r-project.org/doc/manuals/R-intro.pdf

Moodle:
A moodle course is associated with this lecture. Please log into moddle at https://moodle.umass.edu/ and confirm that the course **PUBHLTH690T** is listed under your courses for this semester. Supplemental material for the class will be posted here. The homework sets and final project will be posted on moodle and have to be submitted through moodle.

Grading:
Homework: 60%
Final Project: 40%

Homework:
The homework sets will consist of problem-set-style assignments and implementation exercises. The homework sets will be posted to moodle on the given date. The solutions have to be submitted through moodle on the indicated date by 3:00 PM. Submit a textual answer to each problem. Additionally, for problems that require implementation, a working R-implementation of the solution has to be submitted. Using `RMarkdown` is recommended, and submission of the source file (`.Rmd`) is accepted. Alternatively, you can submit the textual solutions as a pdf-file and the R-implementation in a script-file (`.r`). Collaboration on homework is encouraged, although every student must write up and submit their own assignment (no copy and paste).

Final project:
Students are expected to apply a selection of the methods introduced throughout the course to analyze a given dataset. Students are expected to work on the final project in self-assigned groups of 2-3. The final

project assignment will be posted on Wednesday, April 15. The assignments of students into groups should be communicated to the instructor before that date. The implementations and solutions to the problems (same format as homework) has to be submitted through moodle by midnight on Thursday, May 7.

Course Outline (26 Lectures)

Date	Day	Note	Content
1/19	M	No Class	Martin Luther King Day
1/21	W		Review Syllabus, Brief Introduction to R
1/26	M		Intro to R (cont'd)
1/28	W		1. Genetic Association Studies (Overview, Data Components and Terminology, Data Examples)
2/2	M	Homework 1 posted	
2/4	W		2. Elementary Statistical Principles (Background, Measures and Tests of Association, Analytic Challenges)
2/9	M	Homework 1 due	
2/11	W		
2/16	M	No Class	Presidents' Day
2/17	Tu	Monday class / Homework 2 posted	3. Genetic Data Concepts and Tests (Linkage Disequilibrium, Hardy-Weinberg Equilibrium, Quality Control and Preprocessing)
2/18	W		
2/23	M	Homework 2 due	
2/25	W		
3/2	M	Homework 3 posted	
3/4	W		4. Multiple Comparison Procedures (Measures of Error, Single-step and Step-down Adjustments, Resampling-based Methods, Alternative Paradigms)
3/9	M	Homework 3 due	
3/11	W		
3/16	M	No Class	Spring Break
3/18	W	No Class	Spring Break
3/23	M	Homework 4 posted	4. Multiple Comparison (cont'd)
3/25	W		
3/30	M	Homework 4 due	5. Methods for Unobservable Phase (Haplotype Estimation, Estimating and Testing for Haplotype–Trait Association)
4/1	W		
4/6	M	Homework 5 posted	
4/8	W		6. Classification and Regression Trees (Building a Tree, Optimal Trees)
4/13	M	Homework 5 due	
4/15	W	Final project posted	
4/20	M	No Class	Patriot's Day
4/22	W	Monday class	7. Introduction to Coalescent Theory (The Coalescent, Testing and Inference under the Coalescent)
4/27	M		
4/29	W		
5/4	M	No Class	Final Week
5/6	W	No Class	Final Week
5/7	Th	Final project due	

Academic Honesty Policy Statement:

Since the integrity of the academic enterprise of any institution of higher education requires honesty in scholarship and research, academic honesty is required of all students at the University of Massachusetts Amherst.

Academic dishonesty is prohibited in all programs of the University. Academic dishonesty includes but is not limited to: cheating, fabrication, plagiarism, and facilitating dishonesty. Appropriate sanctions may be imposed on any student who has committed an act of academic dishonesty. Instructors should take reasonable steps to address academic misconduct. Any person who has reason to believe that a student has committed academic dishonesty should bring such information to the attention of the appropriate course instructor as soon as possible. Instances of academic dishonesty not related to a specific course should be brought to the attention of the appropriate department Head or Chair. The procedures outlined below are intended to provide an efficient and orderly process by which action may be taken if it appears that academic dishonesty has occurred and by which students may appeal such actions.

Since students are expected to be familiar with this policy and the commonly accepted standards of academic integrity, ignorance of such standards is not normally sufficient evidence of lack of intent. For more information about what constitutes academic dishonesty, please see the Dean of Students' website: http://umass.edu/dean_students/codeofconduct/acadhonesty/

Disability Statement

The University of Massachusetts Amherst is committed to providing an equal educational opportunity for all students. If you have a documented physical, psychological, or learning disability on file with Disability Services (DS), Learning Disabilities Support Services (LDSS), or Psychological Disabilities Services (PDS), you may be eligible for reasonable academic accommodations to help you succeed in this course. If you have a documented disability that requires an accommodation, please notify me within the first two weeks of the semester so that we may make appropriate arrangements.