PubH 8446-001
Advanced Statistical Genetics and Genomics
Spring 2018

Credits: 3
Meeting Days: M/W
Meeting Time: 11:15 – 12:30
Meeting Place: Moos Health Sci Tower 2-116
Instructor: Baolin Wu, PhD; Cavan Reilly, PhD
Office Phone: 612-624-0647 (Wu); 612-624-9644 (Reilly)
Fax: 612-626-0660
E-mail: baolin@umn.edu, cavanr@biostat.umn.edu
Office Hours: TBD

I. Course Description
Statistical methods for low- and high-level analysis of genetic and genomic data including topics on multiple comparison and gene network modeling. Note this course is a continuation of PubH 8445 for PhD students in Biostatistics or Statistics.

II. Course Prerequisites
PubH 8445 Statistics for Human Genetics and Molecular Biology, Statistical theory at the level of Stat 8101-2, a college-level molecular genetics course is recommended, or permission by instructors.

III. Course Goals and Objectives
After taking the course, the students are expected to understand the statistical challenges in genetics and genomics and important statistical methods developed for this field. The students should then be able to conduct independent reading and research in the future.

IV. Methods of Instruction and Work Expectations
The course will focus on four broad research topics over the semester. An introduction covering general research problems and topics in the field will be given in the first week. Each research topic will be presented in 2-4 week period in the rest of the semester (see class schedule). Students are expected to carefully read all the required readings, and are strongly encouraged to also read the optional readings.

V. Course Text and Readings

No required textbooks. Materials will mainly be drawn from the literature. The readings are required (unless identified as optional). The readings have been carefully chosen from the field to represent very good and recent research papers.

The required and optional readings can be retrieved through the University of Minnesota's e-journals. If you have any difficulty accessing any readings, please contact the instructor.

VI. Course Outline/Weekly Schedule

Week 1-2 Introduction to genome-wide association study (GWAS): association test of common variants, multiple hypothesis testing (FWER)

Week 3-4 Introduction to genome-wide sequencing study: association test of rare variants

Week 4-5 Introduction to analysis of GWAS summary data: empirical Bayes modeling (FDR control), and incorporate functional information

Week 6-7 Introduction to association study of methylation and microbiome data: multiple hypothesis testing (FDR control) and kernel machine modeling

Week 8 Biological Sequence Analysis: dynamic programming and the extreme value distribution

Week 9 Biological Sequence Analysis: hidden Markov models, computational gene recognition

Week 10 Biological Sequence Analysis: feature detection, multiple alignment and motif discovery

Week 11 Introduction to next generation sequencing-applications, e.g. ChIP-seq, variant calls

Week 12 RNA-seq analysis as an alternative to microarrays-low level analysis computational details

Week 13 Computational aspects of next generation sequencing

Week 14 Quantification of signal in mass spectrometry, tandem mass spectrometry and algorithms for tandem mass spectrometry

Week 15 Metabolomics-LC/GC-MS-low level analysis with computational details

VII. Evaluation and Grading

Course grade is based on homeworks (70%) and final project (30%). A letter grade will be determined from the percentage of points each student receives. The curve for final grades will be: A = 95-100; A- = 90-94; B+ = 85-89; B = 80-84; B- = 75-79; C+ = 70-74; C = 65-69; C- = 60-64; F = below 60. For those registered S/N, S = 60-100.

See the course schedule for homework assignment due date. Unless arrangements have been made with the instructor PRIOR to the due date, homework turned in after it is due will have 50% points deducted and no credit will be given after the next homework is due.

The University utilizes plus and minus grading on a 4.000 cumulative grade point scale in accordance with the following:

A 4.000 - Represents achievement that is outstanding relative to the level necessary to meet course requirements

A- 3.667

B+ 3.333
B 3.000 - Represents achievement that is significantly above the level necessary to meet course requirements

B- 2.667

C+ 2.333

C 2.000 - Represents achievement that meets the course requirements in every respect

C- 1.667

D+ 1.333

D 1.000 - Represents achievement that is worthy of credit even though it fails to meet fully the course requirements

S Represents achievement that is satisfactory, which is equivalent to a C- or better.

For additional information, please refer to:
http://policy.umn.edu/Policies/Education/Education/GRADINGTRANSCRIPTS.html

Course Evaluation
The SPH will collect student course evaluations electronically using a software system called CoursEval:
www.sph.umn.edu/courseval. The system will send email notifications to students when they can access and complete their course evaluations. Students who complete their course evaluations promptly will be able to access their final grades just as soon as the faculty member renders the grade in SPHGrades:
www.sph.umn.edu/grades. All students will have access to their final grades through OneStop two weeks after the last day of the semester regardless of whether they completed their course evaluation or not. Student feedback on course content and faculty teaching skills are an important means for improving our work. Please take the time to complete a course evaluation for each of the courses for which you are registered.

Incomplete Contracts
A grade of incomplete “I” shall be assigned at the discretion of the instructor when, due to extraordinary circumstances (e.g., documented illness or hospitalization, death in family, etc.), the student was prevented from completing the work of the course on time. The assignment of an “I” requires that a contract be initiated and completed by the student before the last official day of class, and signed by both the student and instructor. If an incomplete is deemed appropriate by the instructor, the student in consultation with the instructor, will specify the time and manner in which the student will complete course requirements. Extension for completion of the work will not exceed one year (or earlier if designated by the student’s college). For more information and to initiate an incomplete contract, students should go to SPHGrades at:
www.sph.umn.edu/grades.

University of Minnesota Uniform Grading and Transcript Policy
A link to the policy can be found at onestop.umn.edu.

VIII. Other Course Information and Policies

Grade Option Change (if applicable):
For full-semester courses, students may change their grade option, if applicable, through the second week of the semester. Grade option change deadlines for other terms (i.e. summer and half-semester courses) can be found at onestop.umn.edu.

Course Withdrawal:
Students should refer to the Refund and Drop/Add Deadlines for the particular term at onestop.umn.edu for information and deadlines for withdrawing from a course. As a courtesy, students should notify their instructor and, if applicable, advisor of their intent to withdraw.
Students wishing to withdraw from a course after the noted final deadline for a particular term must contact the School of Public Health Office of Admissions and Student Resources at sph-ssc@umn.edu for further information.

**Student Conduct Code:**
The University seeks an environment that promotes academic achievement and integrity, that is protective of free inquiry, and that serves the educational mission of the University. Similarly, the University seeks a community that is free from violence, threats, and intimidation; that is respectful of the rights, opportunities, and welfare of students, faculty, staff, and guests of the University; and that does not threaten the physical or mental health or safety of members of the University community.

As a student at the University you are expected adhere to Board of Regents Policy: Student Conduct Code. To review the Student Conduct Code, please see: http://regents.umn.edu/sites/default/files/policies/Student_Conduct_Code.pdf.

Note that the conduct code specifically addresses disruptive classroom conduct, which means “engaging in behavior that substantially or repeatedly interrupts either the instructor's ability to teach or student learning. The classroom extends to any setting where a student is engaged in work toward academic credit or satisfaction of program-based requirements or related activities.”

**Use of Personal Electronic Devices in the Classroom:**
Using personal electronic devices in the classroom setting can hinder instruction and learning, not only for the student using the device but also for other students in the class. To this end, the University establishes the right of each faculty member to determine if and how personal electronic devices are allowed to be used in the classroom. For complete information, please reference: http://policy.umn.edu/Policies/Education/Education/STUDENTRESP.html.

**Scholastic Dishonesty:**
You are expected to do your own academic work and cite sources as necessary. Failing to do so is scholastic dishonesty. Scholastic dishonesty means plagiarizing; cheating on assignments or examinations; engaging in unauthorized collaboration on academic work; taking, acquiring, or using test materials without faculty permission; submitting false or incomplete records of academic achievement; acting alone or in cooperation with another to falsify records or to obtain dishonestly grades, honors, awards, or professional endorsement; altering, forging, or misusing a University academic record; or fabricating or falsifying data, research procedures, or data analysis. (Student Conduct Code: http://regents.umn.edu/sites/default/files/policies/Student_Conduct_Code.pdf) If it is determined that a student has cheated, he or she may be given an "F" or an "N" for the course, and may face additional sanctions from the University. For additional information, please see: http://policy.umn.edu/Policies/Education/Education/INSTRUCTORRESP.html.

The Office for Student Conduct and Academic Integrity has compiled a useful list of Frequently Asked Questions pertaining to scholastic dishonesty: http://www1.umn.edu/oscai/integrity/student/index.html. If you have additional questions, please clarify with your instructor for the course. Your instructor can respond to your specific questions regarding what would constitute scholastic dishonesty in the context of a particular class-e.g., whether collaboration on assignments is permitted, requirements and methods for citing sources, if electronic aids are permitted or prohibited during an exam.

**Makeup Work for Legitimate Absences:**
Students will not be penalized for absence during the semester due to unavoidable or legitimate circumstances. Such circumstances include verified illness, participation in intercollegiate athletic events, subpoenas, jury duty, military service, bereavement, and religious observances. Such circumstances do not include voting in local, state, or national elections. For complete information, please see: http://policy.umn.edu/Policies/Education/Education/MAKEUPWORK.html.

**Appropriate Student Use of Class Notes and Course Materials:**
Taking notes is a means of recording information but more importantly of personally absorbing and integrating the educational experience. However, broadly disseminating class notes beyond the classroom community or accepting compensation for taking and distributing classroom notes undermines instructor interests in their intellectual work product while not substantially furthering instructor and student interests in effective learning. Such actions violate shared norms and standards of the academic community. For additional information, please see: http://policy.umn.edu/Policies/Education/Education/STUDENTRESP.html.
Sexual Harassment:
“Sexual harassment” means unwelcome sexual advances, requests for sexual favors, and/or other verbal or physical conduct of a sexual nature. Such conduct has the purpose or effect of unreasonably interfering with an individual's work or academic performance or creating an intimidating, hostile, or offensive working or academic environment in any University activity or program. Such behavior is not acceptable in the University setting. For additional information, please consult Board of Regents Policy: http://regents.umn.edu/sites/default/files/policies/SexHarassment.pdf

Equity, Diversity, Equal Opportunity, and Affirmative Action:
The University will provide equal access to and opportunity in its programs and facilities, without regard to race, color, creed, religion, national origin, gender, age, marital status, disability, public assistance status, veteran status, sexual orientation, gender identity, or gender expression. For more information, please consult Board of Regents Policy: http://regents.umn.edu/sites/default/files/policies/Equity_DiversityEO-AA.pdf

Disability Accommodations:
The University of Minnesota is committed to providing equitable access to learning opportunities for all students. The Disability Resource Center Student Services is the campus office that collaborates with students who have disabilities to provide and/or arrange reasonable accommodations.

If you have, or think you may have, a disability (e.g., mental health, attentional, learning, chronic health, sensory, or physical), please contact DRC at 612-626-1333 or drc@umn.edu to arrange a confidential discussion regarding equitable access and reasonable accommodations.

If you are registered with DS and have a current letter requesting reasonable accommodations, please contact your instructor as early in the semester as possible to discuss how the accommodations will be applied in the course.

For more information, please see the DS website, https://diversity.umn.edu/disability/.

Mental Health and Stress Management:
As a student you may experience a range of issues that can cause barriers to learning, such as strained relationships, increased anxiety, alcohol/drug problems, feeling down, difficulty concentrating and/or lack of motivation. These mental health concerns or stressful events may lead to diminished academic performance and may reduce your ability to participate in daily activities. University of Minnesota services are available to assist you. You can learn more about the broad range of confidential mental health services available on campus via the Student Mental Health Website: http://www.mentalhealth.umn.edu.

The Office of Student Affairs at the University of Minnesota:
The Office for Student Affairs provides services, programs, and facilities that advance student success, inspire students to make life-long positive contributions to society, promote an inclusive environment, and enrich the University of Minnesota community.
Units within the Office for Student Affairs include, the Aurora Center for Advocacy & Education, Boynton Health Service, Central Career Initiatives (CCE, CDes, CFANS), Leadership Education and Development – Undergraduate Programs (LEAD-UP), the Office for Fraternity and Sorority Life, the Office for Student Conduct and Academic Integrity, the Office for Student Engagement, the Parent Program, Recreational Sports, Student and Community Relations, the Student Conflict Resolution Center, the Student Parent HELP Center, Student Unions & Activities, University Counseling & Consulting Services, and University Student Legal Service.

For more information, please see the Office of Student Affairs at http://www.osa.umn.edu/index.html.

Academic Freedom and Responsibility: for courses that do not involve students in research:
Academic freedom is a cornerstone of the University. Within the scope and content of the course as defined by the instructor, it includes the freedom to discuss relevant matters in the classroom. Along with this freedom comes responsibility. Students are encouraged to develop the capacity for critical judgment and to engage in a sustained and independent search for truth. Students are free to take reasoned exception to the views offered in any course of study and to reserve judgment about matters of opinion, but they are responsible for learning the content of any course of study for which they are enrolled.*
OR:

**Academic Freedom and Responsibility, for courses that involve students in research**

Academic freedom is a cornerstone of the University. Within the scope and content of the course as defined by the instructor, it includes the freedom to discuss relevant matters in the classroom and conduct relevant research. Along with this freedom comes responsibility. Students are encouraged to develop the capacity for critical judgment and to engage in a sustained and independent search for truth. Students are free to take reasoned exception to the views offered in any course of study and to reserve judgment about matters of opinion, but they are responsible for learning the content of any course of study for which they are enrolled.* When conducting research, pertinent institutional approvals must be obtained and the research must be consistent with University policies.

Reports of concerns about academic freedom are taken seriously, and there are individuals and offices available for help. Contact the instructor, the Department Chair, your adviser, the associate dean of the college, (Dr Kristin Anderson, SPH Dean of Student Affairs), or the Vice Provost for Faculty and Academic Affairs in the Office of the Provost.

* Language adapted from the American Association of University Professors "Joint Statement on Rights and Freedoms of Students".

**Student Academic Success Services (SASS):** [http://www.sass.umn.edu](http://www.sass.umn.edu)

Students who wish to improve their academic performance may find assistance from Student Academic Support Services. While tutoring and advising are not offered, SASS provides resources such as individual consultations, workshops, and self-help materials.

*Template update 9/2014*