

**PubH 7445**  
**Statistics for Human Genetics and Molecular Biology**  
**Fall 2017**

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**Credits:** 3  
**Meeting Days:** MWF  
**Meeting Time:** 1:25-2:15  
**Meeting Place:** Mayo C381  
**Instructor:** Cavan Reilly PhD  
**Office Address:** 359 University Office Plaza  
**Office Phone:** 612-624-9644  
**Fax:** 612-626-0660  
**E-mail:** [cavanr@biostat.umn.edu](mailto:cavanr@biostat.umn.edu)  
**Office Hours:** MW 2:30-3:30

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**I. Course Description**

This master's-level course focuses on quantitative skills and knowledge for interdisciplinary applications in genetics and molecular biology. Topics include gene association mapping, analyses of gene expression data from microarray experiments, analysis of next generation sequence data, false discovery rate estimation, gene set enrichment analysis and machine learning algorithms. Students will conduct extensive data analysis using R/Bioconductor.

**Acknowledgments**

The contents of PubH 7445 have been developed with the contributions of numerous instructors. Dr. Reilly, the current instructor, has been involved with the majority of recent content and modifications. Former faculty/instructors, including Drs. Yen-Yi Ho, Wei Pan and Hegang Chen, all had roles in either the conceptual development or actual content of the current course, and are acknowledged for their contributions.

**II. Course Prerequisites**

Pub 6450-6451 or equivalent or permission of instructor. Some background with molecular biology is desirable but not required.

**III. Course Goals and Objectives**

By the end of the course, students should have a basic knowledge of statistical tools applied to human genetics, basic R/Bioconductor programming skills and substantial experience applying these skills and experience interpreting results and communicating these results to others.

#### **IV. Methods of Instruction and Work Expectations**

The course will use lectures and in class structured learning experiences to facilitate learning. Students will be evaluated based on their performance on homework exercises and a final project.

#### **V. Course Text and Readings**

John Verzani's SimpleR notes (<http://cran.r-project.org/doc/contrib/VerzaniSimpleR.pdf>)

Foulkes (2009): Applied Statistical Genetics with R

Hahne, Huber, Gentleman, and Falcon (2008): Bioconductor Case Studies

#### **VI. Course Outline/Weekly Schedule**

Week 1: Introduction/review of basic concepts of genetics and molecular biology

Week 2: Introduction/review of basic statistics and the use of R

Week 3: Linkage disequilibrium and Hardy Weinberg equilibrium

Week 4: Multiple comparisons

Week 5: Haplotype estimation

Week 6: Classification/supervised learning with categorical predictors

Week 7: More on classification/supervised learning

Week 8: Introduction to microarray data analysis

Week 9: Inference for differential expression

Week 10: More on inference for differential expression

Week 11: Next generation sequencing: bioinformatics

Week 12: Next generation sequencing: biostatistics

Week 13: Machine learning approaches to gene expression data

Week 14: R tools for pathway analysis in RNA-Seq data

Week 15: Proteomics and metabolomics

#### **VII. Evaluation and Grading**

Performance on homework assignments (70%) and the final course project (30%).

Late homework will lose 30% of the total points each day it is late, unless prior arrangements have been made. Homework will not be accepted after the time at which graded homework are returned.

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 CourseEval: [www.sph.umn.edu/courseeval](http://www.sph.umn.edu/courseeval). 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](http://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](http://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](http://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](http://onestop.umn.edu).

**Course Withdrawal:**

Students should refer to the Refund and Drop/Add Deadlines for the particular term at [onestop.umn.edu](http://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](mailto: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](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](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\\_Diversity\\_EO\\_AA.pdf](http://regents.umn.edu/sites/default/files/policies/Equity_Diversity_EO_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](mailto: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>:**

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.