

HUGEN 2029: Introduction to Gene Mapping
3 credits / Fall Term 2019 / Mondays and Wednesdays, 9:30-10:55 am / 3121C Public Health

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Office Hours: By appointment

Faculty Availability: I welcome your questions and suggestions. Please feel free to set up an appointment. Also, if you are having any problems with the course, please contact me as soon as possible. E-mail is a good way to reach me. However, since I get many e-mails, please use an informative subject line starting with "HUGEN 2029".

Course Description: This course presents a literature-based approach to understanding and interpreting results from gene mapping papers in the field of human genetics. Traditional and state-of-the-art genetic mapping methodologies will be explored.

Course Overview and Learning Objectives:

This course covers the (currently) most commonly-used technologies and methodologies for discovering and exploring genotype-phenotype associations. Each methodology will be covered in one or two didactic class sessions, and then participants will read, critique, and present papers that apply the methodology.

At the end of this course, participants should be able to:

- Describe the mathematical and scientific underpinnings of each methodology
- Discuss how the choice of study design influences the choice of methodology (and vice versa)
- Discuss the strengths and limitations of each methodology
- Evaluate gene mapping results in the current literature
- Critique the study design and methodology choices in published gene-mapping studies

Texts/assigned materials:

Participants will need to read and be prepared to discuss assigned materials that will be posted for personal use on CourseWeb (Blackboard). There is no required textbook for this course.

Exams and Assignments:

- **Exams:** There will be two (take-home) exams, a mid-term exam and a final exam, to assess students' ability to understand and critique gene mapping methods. The format will be open-ended questions.

- **Student presentations:** Each student will review, present and discuss a (recently) published paper in class. Presentations should be short, approximately 20 minutes, and primarily be an introduction to the topic and briefly highlight the paper. The student will subsequently lead a discussion of the paper. The goal of these presentations is to learn to critically review papers, evaluate the strengths and weaknesses of the papers, and to gain experience in public speaking. Everyone is expected to have read the selected papers in advance and come prepared to discuss.

- **Discussions:** In addition to the student presentations, the class as a whole will review and discuss several papers. Everyone is expected to have read the selected papers in advance and come prepared to discuss.

Student Performance Evaluation:

All course requirements must be completed to receive credit for the course. Evaluation will be based on the following components:

- Attendance and Quality of Contribution to Discussion (25% of final grade)

Attendance, active participation in class discussions, and evidence of being prepared for class (including having read the assigned readings and completion of assignments) are expected. While cell phones and laptops/tablets may be used to access slides or assigned readings, take notes, etc., please do not use them during class time for non-class purposes. If you will miss a class, please let me know in advance.

- Student Presentations (25% of final grade)

- Mid-term Exam (25% of final grade)

Take-home mid-term exam will be posted on CourseWeb on October 16. The mid-term exam is due on October 23 (by midnight; please submit to: bbd3@pitt.edu).

- Final Exam (25% of final grade)

Take-home final exam will be posted on CourseWeb on December 4. The final exam is due on December 11 (by midnight; please submit to: bbd3@pitt.edu).

Grade option:	Grading scale:		
Letter grade	97 - 100% A+	77 – 79.9% C+	< 60% F
	93 – 96.9% A	73 – 76.9% C	
	90 – 92.9% A-	70 – 72.9% C-	
	87 – 89.9% B+	67 – 69.9% D+	
	83 – 86.9% B	63 – 66.9% D	
	80 – 82.9% B-	60 – 62.9% D-	

CourseWeb/BlackBoard:

The University's CourseWeb (BlackBoard) will be used to post announcements, assignments, and readings for personal use.

Course Policies:

The Internet generally should not be accessed during class, except to access course slides or assigned readings, help resolve a disputed point in discussion or otherwise enhance discussion. Students should check their email regularly to ensure that they receive relevant communication regarding the course.

Students should familiarize themselves also with the following policies:

- Academic Integrity Policy:

All individuals (students, faculty, post-doctoral researchers, and staff) at Pitt Public Health abide by the University's policy on academic integrity. In accordance with this policy, the school maintains an outline of the procedural sequence of events to occur when violations of academic integrity are brought to the attention of administrative leaders. The full policy is available in the Academic Handbook.

All students are expected to adhere to the school's standards of academic honesty. Cheating/plagiarism will not be tolerated. The Graduate School of Public Health's policy on academic integrity, approved by EPCC on 7/31/18, which is based on the University policy, is available online in the Pitt Public Health Academic Handbook www.publichealth.pitt.edu/home/academics/academic-requirements. The policy includes obligations for faculty and students, procedures for adjudicating violations, and other critical information. Please take the time to read this policy.

Students should be especially mindful of guidelines on academic integrity and take care to avoid plagiarizing the work - including the ideas or words - of their colleagues (fellow course participants) or other authors.

Students are encouraged to discuss their ideas and work together; however, a citation to a fellow student should be provided when appropriate.

- Diversity and Academic Civility Statement:

In this course, students, faculty and guests represent a diversity of individual perspectives, backgrounds, and experiences, which enriches our classes. We urge all to be respectful of others.

The University of Pittsburgh Graduate School of Public Health considers the diversity of its students, faculty, and staff to be a strength and critical to its educational mission. Pitt Public Health is committed to creating and fostering inclusive learning environments that value human dignity and equity. Every member of our community is expected to be respectful of the individual perspectives, experiences, behaviors, worldviews, and backgrounds of others. While intellectual disagreement may be constructive, no derogatory statements, or demeaning or discriminatory behavior will be permitted.

If you feel uncomfortable or would like to discuss a situation, please contact any of the following:

- the course instructors;
- the Pitt Public Health Associate Dean for Diversity at 412-624-3506 or nam137@pitt.edu;
- the University's Office of Diversity and Inclusion at 412-648-7860 or <https://www.diversity.pitt.edu/make-report/report-form> (anonymous reporting form)

- Accommodation for Students with Disabilities:

If you have any disability for which you may require accommodation, you are encouraged to notify both your instructor and the Office of Disability Resources and Services (DRS), 140 William Pitt Union (Voice or TTD 412-648-7890), <http://www.studentaffairs.pitt.edu/drs/>, drsrecep@pitt.edu, as early as possible in the term. Information on applying for services can be found here: <http://www.studentaffairs.pitt.edu/drs/students/apply/>

- Accessibility:

Blackboard is ADA Compliant and has fully implemented the final accessibility standards for electronic and information technology covered by Section 508 of the Rehabilitation Act Amendments of 1998. Please note that, due to the flexibility provided in this product, it is possible for some material to inadvertently fall outside of these guidelines.

- Copyright Notice:

Course materials may be protected by copyright. United States copyright law, 17 USC section 101, et seq., in addition to University policy and procedures, prohibit unauthorized duplication or retransmission of course materials. See [Library of Congress Copyright Office](#) and the [University Copyright Policy](#).

- Classroom Recording:

To ensure the free and open discussion of ideas, students may not record classroom lectures, discussion and/or activities without the advance permission of the instructors, and any such recording properly approved in advance can be used solely for the student's own private use or for all students enrolled in this class only but may not be further copied, distributed, published, or otherwise used for any other purpose without the express written consent of the course instructors. Any student who records a class session must provide a copy of the recording to the instructors if requested to do so.

- Sexual Misconduct, Required Reporting, and Title IX:

The University is committed to combatting sexual misconduct. As a result, you should know that University faculty and staff members are required to report any instances of sexual misconduct, including harassment and sexual violence, to the University's Title IX office so that the victim may be provided appropriate resources and support options. What this means is that as your professors, we are required to report any incidents of sexual misconduct that are directly reported to us, or of which we are somehow made aware.

There are two important exceptions to this requirement about which you should be aware:

- (1) A list of the designated University employees who, as counselors and medical professionals, do not have this reporting responsibility and can maintain confidentiality, can be found here: www.titleix.pitt.edu/report/confidentiality

(2) An important exception to the reporting requirement exists for academic work. Disclosures about sexual misconduct that are shared as part of an academic project, classroom discussion, or course assignment, are not required to be disclosed to the University's Title IX office

If you are the victim of sexual misconduct, Pitt encourages you to reach out to these resources:

- Title IX Office: 412-648-7860 (<https://www.titleix.pitt.edu/>)
- SHARE @ the University Counseling Center: 412-648-7930 (8:30 A.M. TO 5 P.M. M-F) and 412-648-7856 (AFTER BUSINESS HOURS)

If you have a safety concern, please contact the University of Pittsburgh Police, 412-624-2121.

Other reporting information is available here: <https://www.titleix.pitt.edu/resources>

HUGEN 2029: Introduction to Gene Mapping
Fall Term 2019 Schedule (August 19, 2019)
(Mondays and Wednesdays, 9:30-10:55 am / 3121C Public Health)

Date	Topics
Monday August 26	Course introduction
Wednesday August 28	Study design, study populations, diversity and sex
Monday September 2	LABOR DAY HOLIDAY – No class
Wednesday September 4	Candidate gene association studies
Monday September 9	Genome-wide association studies (GWAS)
Wednesday September 11	GWAS – imputation and combining datasets
Monday September 16	GWAS – polygenic risk scores (and what about the environment?)
Wednesday September 18	GWAS discussion
Monday September 23	GWAS follow-up; eQTL analysis, gene set analysis
Wednesday September 25	GWAS follow-up discussion
Monday September 30	Sequencing - biochemistry, rare variants, and cancer
Wednesday October 2	Sequencing - association studies
Monday October 7	Sequencing discussion
Wednesday October 9	Family-based designs, linkage analysis
Monday October 14	Family-based studies discussion
Wednesday October 16	No class – mid-term exam posted on CourseWeb
Monday October 21	No class
Wednesday October 23	No class – mid-term exam due (by midnight)
Monday October 28	Family-based studies discussion
Wednesday October 30	Family-based studies discussion
Monday November 4	Epigenetics, methylation
Wednesday November 6	Expression data, transcriptome-wide association studies, RNA-seq, single cell RNA-seq
Monday November 11	Expression discussion
Wednesday November 13	Copy number variants (CNVs) / role of CNVs in disease
Monday November 18	Post-transcriptional regulation
Wednesday November 20	Other genomes: the microbiome and mitochondria
Monday November 25/28	THANKSGIVING HOLIDAY – No class
Monday December 2	Multi-omics approaches
Wednesday December 4	Mendelian randomization – final exam posted on CourseWeb
Monday December 9	Topic tbd
Wednesday December 11	No class – final exam due (by midnight)