EPI 507: Principles of Genetic Epidemiology

Fall 2 2026

Meeting Time: Monday/Wednesday 09:45 - 11:15 AM
Location: FXB G13 (HSPH)
Credits: 2.5 credits
Website: https://canvas.harvard.edu/courses/159480
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Instructor Information

Faculty

Dan Chasman, PhD
Associate Professor, Medicine, Harvard Medical School
Associate Professor, Department of Epidemiology, Harvard T.H. Chan School of Public Health
Associate Geneticist, Department of Medicine, Brigham and Women's Hospital
Email: dchasman@bwh.harvard.edu
Office hours: Tuesday 1-2 pm or by appointment

Jie (Jack) Hu, MD, PhD
Instructor, Center for Genomic Medicine, Massachusetts General Hospital
Instructor, Department of Epidemiology, Harvard T.H. Chan School of Public Health
Email: Jie.Hu@mgh.harvard.edu
Office hours: Wednesday 2-3 pm or by appointment

Teaching Fellows

Buu Truong, MD
Email: btruong@hsph.harvard.edu
Office Hours: Thursday, 4-5 pm or by appointment

Avon Yang, MS
Email: avon_yang@fas.harvard.edu
Office hours: TBD or by appointment

(Please see canvas Teaching. Staff page for Zoom links.)

Course Description

Genetic epidemiology is the study of genetic influences on human traits and disease in populations with direct clinical and public health applications. Genetic epidemiology also contributes to a deeper understanding of disease mechanisms, such as disease biology, etiology, and pathophysiology, all potentially informing development of new therapeutics. This course introduces the goals and methods of genetic epidemiology. After a review of the history of genetic epidemiology and basic molecular genetics, the course will cover study designs, data resources, and analytic approaches for related but distinct goals, including: genome-wide association studies (GWAS) and gene discovery; clinical and public health applications of genetics; and understanding the joint contributions of inherited genetics, social factors, and environmental exposures to disease etiology and the distribution of disease in populations. The course will also review empirical findings on the contribution of genetics to the understanding of human traits diseases with some applications in healthcare.

The target audience includes doctoral and masters' students looking for a non-specialist introduction to genetic epidemiology. This course is required for epidemiology Ph.D. students and fulfills a core requirement for Computational Biology and Quantitative Genomics M.Sc. students. Pre-Requisites: [EPI 201 and (EPI 202 or BST 210 or BST 220)] or PHS 2000A or BST 213.

Learning Objectives

Upon successful completion of this course, students will be able to:

- Describe how molecular, Mendelian, and population genetics relate to epidemiology
- Use publicly available resources describing genetic variation in human populations and its association with diseases and traits
- Relate concepts from genetic epidemiology to general epidemiologic concepts (e.g. population stratification bias is an example of confounding)
- Evaluate and develop appropriate and well-powered study designs and analytic approaches for understanding the relationship between genetic variation and variation in human traits
- Critically evaluate genetic association studies as well as claims regarding the importance of genetic variation in different contexts (basic research, clinical epidemiology, public health)
- Distinguish concepts of race/ethnicity from genetic ancestry and understand how to appropriately incorporate race/ethnicity/ancestry in genetic studies

Competencies

This course covers the following Program/Field of Study Competencies:

SM 80 CBQG

1. Examine the basics of techniques modern population genetics such as genome wide association study methodology

Course Structure

This course will consist of lectures, in-class interactive exercises and discussions (including small group activities), individual homeworks, asynchronous recorded vignettes, online exercises (Canvas quizzes), and a final group project (see below). The lectures will not be offered via Zoom. Lectures will be recorded but made available only for exceptional circumstances.

The Canvas site is an important learning tool for this course where students will access required articles and recordings, submit course assignments, and share other resources with the class. Students will be expected to check the Canvas site on a weekly basis for course announcements.

During the first two weeks (until the drop date), students will assemble into small groups, targeting 4 students per group, for the final project and interactive activities throughout the course. Students are expected to participate in discussions and exercises in class and are encouraged, but not required, to do so within their group.

Course Readings and Pre-recorded Vignettes

There is no required textbook for this class. Readings will consist of book chapters, journal articles, recorded didactic video vignettes available through PDFs and URL links supplied via the Canvas site. Required readings for each week are listed on the Canvas site. The vignettes were prepared during the COVID-19 pandemic to convey the foundational concepts of genetic epidemiology and complement the readings. Lecture material, readings, and the vignettes form the basis of quizzes, homeworks, and in-class discussions.

Grading, Progress, and Assessment

The final grade for this course will be based on:

- Canvas Quizzes (10%)

- Team-learning exercises (15%)
- Homework (40%)
- Final project (35%)
- New this year: Optional bonus (5%) in addition to preceding

Canvas Quizzes (10%)

The canvas quizzes will be graded on a participation basis**.** That is, full credit for timely completion, regardless of whether answers are correct. Note that the quiz questions are designed to be helpful for checking understanding of the course material and for completing homework.

Team-learning exercises (15%)

In-class exercises will be graded on a participation basis and completed, preferably and most efficiently within groups, although working within groups is not required**.** These exercises will cover hands-on skills in genetic data resources and analytic approaches relevant to the 1) UCSC Genome Browser, 2) LD link, 3) GWAS Catalog, and 4) gnomAD. The materials for in-class exercises will be available on the Canvas site. There will be 4 in-class exercises throughout the semester on the following dates.

Exercises #1: Wednesday, November 4 (UCSC browser)
Exercises #2: Monday, November 9 (LD link)
Exercises #3: Wednesday, November 18 (GWAS catalog)
Exercises #4: Monday, December 7 (gnomAD)

Homework (40%)

There will be a total of six graded homeworks throughout the course**.** The homeworks will elaborate on concepts presented in lectures, vignettes, and in-class discussions and provide opportunities to learn and use some of the online resources and software tools introduced in lectures and exercises. It is allowed to discuss the homework assignments with other students (and there will be discussion forum for each homework to the Canvas site), but students must submit their own answers. See also guidance about academic integrity below.

Homeworks will be typically assigned on Wednesdays and due the following **Wednesday before class (before 9:45 AM Boston time**) except 1) homework #3 given 5 extra days to accommodate Veterans Day and 2) homework #4 is given an extra week and due after the Thanksgiving break. Note, the project draft is due on the Wednesday before Thanksgiving (next section). Homeworks should be uploaded to Canvas. The schedule below summarizes the exact due dates for the homeworks.

#	Topic	Assign	Due
1	DNA Sequence, Genetic Variation	W. Oct. 28	W. Nov 4
2	Population Genetics, Genotype/Allele Frequencies and Hardy-Weinberg Equilibrium, UCSC Genome Browser	W. Nov 4	Th. Nov 12
3	Linkage disequilibrium	W. Nov 11	M. Nov 23
4	GWAS basics, GWAS Catalog, Association analysis	W. Nov 18	W. Dec 2
5	Polygenic Risk Scores, Mendelian Randomization, Gene-environment interaction effects	W. Dec 2	W. Dec 9
6	Sequencing, Rare variation, and gnomAD,	W. Dec 9	W. Dec 16

Final project (35%: 5% for draft version, 30% for final version)

Each group will submit a final project. The final project will consist of a short (up to four pages) **proposal** for analysis using available (i.e. public or by application) data sources (e.g. the UK Biobank, TOPMed, dbGaP, All of Us, publicly available GWAS summary statistics, etc.) and a short (<5 mins) “elevator pitch” (in PowerPoint or PDF) presentation describing the proposal. This project will allow students to identify specific hypotheses that they can investigate using genetic epidemiology and choose designs and analyses appropriate for those hypotheses. The project is restricted to publicly available data sets to conform to real-world limitations on technology and budgets and to give students familiarity with available resources. The proposal will consist of the following four sections:

1. Background & Significance
2. Design
3. Analytic Approach
4. Strengths and Limitations

The final project is a group project; most groups will have four (4) members. It is intended that each group member will be primarily responsible for drafting one of the four sections as decided within the group and that the group will work together to harmonize the separate sections. The project will be graded in two phases: the **draft** version, due November 25, 2026 (11:59 PM), and the **final** version, due December 16, 2026 (9:45 AM). All students in a group will receive the 5% credit for submitting the initial draft and meeting with instructors to review comments (below) and the same grade for the final version of the project according to the rubric for the remaining 30% credit. **N.B. Due to time, budget, and computing restraints, the final project is solely an analysis proposal. Students should not actually perform their proposed analyses.**

The instructors will return feedback comments on the draft by December 2, 2026. Between Wednesday December 2 and Friday December 11, the teaching staff will schedule meetings with each group in person or on zoom to review the preparation of the final project. To receive the full 5% participation credit for the draft, all students from a group are expected to attend, unless specifically excused. The final class (Wednesday, December 16, 2026) will be dedicated to brief, i.e. <5 minutes, oral group presentations from each group of their final project with slides (think elevator pitch, e.g. ~1 slide for each section). From the experience of past editions of the course, it cannot be stressed enough that the more developed the ideas in the draft proposal, the better the final project. Students are strongly encouraged to start discussing the project within their groups at the beginning of the term and to reach out to the instructors during office hours for feedback as they develop proposals. Having a clear sense of the aims and analytic approach for the project proposal as early in the term as possible will make the proposal more coherent and also easier to prepare.

Bonus questions on the reading (an additional 5%)

The required (and recommended) readings are highly curated, not too long, and excellent references for the essential material in the course. To encourage students to spend time reading these articles, students may submit a thoughtful response to optional bonus questions through Canvas that accompany the reading. These bonus assignments will be recognized by up to 5% credit in addition to credit for the required graded elements. There are 10 of these questions in total, each worth 0.5%. Responses are expected to be brief, i.e. 1 or 2 sentences, but should reveal decent comprehension of the reading. Any reasonable effort to respond to a question as addressed by the reading material will receive credit. The responses should also reflect the student's own thinking and will be screened for AI generated text. Once the required elements for the course have been scored (above) and score thresholds for letter grades have been determined, each student's accumulated bonus points will be added to their score on the required elements. Thus, doing the reading not only will help master the course material but also has the potential to lift a letter grade for the course, e.g. from A- to A.

Class Attendance

Attendance in-person is expected. The lectures will not be offered for remote learning via Zoom. However, the course staff and HSPH recognize that some absences may be unavoidable. If you cannot attend class for an unavoidable reason, please contact the teaching staff as soon as you anticipate being absent so we can make alternate arrangements.

Late completion of assignments

Late assignments will be graded but will not receive full credit. Excepting unavoidable lateness, deductions of credit will depend on how long after the deadline assignments are submitted, imposing 10% deduction for each day late.

Harvard Chan Policies and Expectations

Inclusivity Statement

Diversity and inclusiveness are fundamental to public health education and practice. Students are encouraged to have an open mind and respect differences of all kinds. The EPI507 teaching team shares responsibility with

you for creating a learning climate that is hospitable to all perspectives and cultures; please feel free to contact us if you have any concerns or suggestions.

Bias Related Incident Reporting

The Harvard T.H. Chan School of Public Health believes all members of our community should be able to study and work in an environment where they feel safe and respected. As a mechanism to promote an inclusive community, we have created an anonymous bias-related incident reporting system. If you have experienced bias, please submit a report here so that the administration can track and address concerns as they arise and to better support members of the Harvard Chan community.

Title IX

The following policy applies to all Harvard University students, faculty, staff, appointees, or third parties: Harvard University Sexual and Gender-Based Harassment Policy. Procedures For Complaints Against a Faculty Member

Procedures For Complaints Against Non-Faculty Academic Appointees

Non-Attribution Community Norm

Per guidance Harvard T.H. Chan School of Public Health, our classroom will incorporate and adopt a community norm of Non-Attribution, modeled after the Chatham House Rule, permitting participants to mention the information discussed in class outside of class but prohibiting attribution of comments or ideas to a specific student, unless that person gives written or verbal permission. This policy is consistent with striving to foster an environment at HSPH grounded in trust, mutual respect, diversity of viewpoints, free expression, open inquiry, and constructive dialogue. To support the highest levels of learning and critical thinking in our classrooms, everyone must feel comfortable sharing their perspectives, ideas, and lived experiences via respectful discussion of complex, sensitive and consequential public health questions and topics.

Note that the course implements the discussion/message board feature within Canvas to provide a forum for exchanges about the homeworks. Entries to the discussion board will typically include an author's name, i.e. attribution. They are further likely to be made when students are not in the classroom, i.e. outside of class, and could potentially violate the Non-Attribution norm. In order that these exchanges comply with the Non-Attribution norm, comments posted to the discussion board should be viewed as equivalent to "in-class" discussions and further strictly focused on the homework questions.

Similarly, some office hour sessions may be attended by more than one student or a group. The Non-Attribution community norm should also apply to this setting. Discussions in office hours should be treated as if they were "in-class." Accordingly, content from office hours should not be attributed to specific students outside of class, although it may be discussed without attribution.

Academic Integrity

Each student in this course is expected to abide by the Harvard University and the Harvard T.H. Chan School of Public Health School's standards of Academic Integrity. All work submitted to meet course requirements is expected to reflect each student's effort as prescribed in the assignment. For example, students must submit their own answers to quizzes and homeworks but answers may be influenced by discussions with other students. Similarly, all students within a group are expected to contribute equally to the entire group project, including drafting, finalizing, and presenting. In the preparation of work submitted to meet course requirements, students should always take great care to distinguish their own ideas and knowledge from information derived from sources.

Students must assume that collaborations, i.e., submitting responses collectively, in the completion of course assignments are prohibited, with exceptions for in-class exercises and the final group project. Students must acknowledge any collaboration and its extent in all submitted work. This requirement applies to collaboration on editing as well as collaboration on substance. Accordingly, submission of homework answers, text for the project, and/or material for the final oral presentations created by generative artificial intelligence (GAI) is not

permitted. However, GAI may be used for help with brainstorming the project. Students who have questions about the use of GAI for assignments or would like to propose a use not addressed here should contact the instructors to discuss permission in advance of such uses.

Should academic misconduct occur, the student(s) may be subject to disciplinary action as outlined in the Student Handbook. See the Student Handbook for additional policies related to academic integrity and disciplinary actions.

Accommodations for Students with Disabilities

Harvard University provides academic accommodations to students with disabilities. Any requests for academic accommodations should ideally be made before the first week of the semester, except for unusual circumstances, so arrangements can be made. Students must register with the Local Disability Coordinator in the Office for Student Affairs to verify their eligibility for appropriate accommodations. Contact Colleen Cronin ccronin@hsph.harvard.edu in all cases, including temporary disabilities.

Religious Holidays, Absence Due to

According to Chapter 151c, Section 2B, of the General Laws of Massachusetts, any student in an educational or vocational training institution, other than a religious or denominational training institution, who is unable, because of his or her religious beliefs, to attend classes or to participate in any examination, study, or work requirement on a particular day shall be excused from any such examination or requirement which he or she may have missed because of such absence on any particular day, provided that such makeup examination or work shall not create an unreasonable burden upon the School. See the student handbook for more information.

Course Evaluations

Constructive feedback from students is a valuable resource for improving teaching. The feedback should be specific, focused and respectful. It should also address aspects of the course and teaching that are positive as well as those which need improvement.

Completion of the evaluation is a requirement for each course. Your grade will not be available until you submit the evaluation. In addition, registration for future terms will be blocked until you have completed evaluations for courses in prior terms.

Course Overview

Week	Lecture		In-class exercise/discussion	Assignment(s) due
	Monday	Wednesday		
1	Oct 26 (DC) - Introduction - Scope of genetic epidemiology	Oct 28 (DC/JH) - Review basic molecular genetics - Groups/project discussion		Oct 28, 9:00 AM (pre-class) Enrollment questionnaire Oct 28, 9:45AM Quiz #1-3
2	Nov 2 (JH) - Mendelian genetics, familial aggregation, heritability	Nov 4 (JH) - Population genetics	Nov 4 (in-class exercise 1, AY) UCSC Genome Browser	Nov 2, 9:45AM Quiz #4-5 Nov 4, 9:45AM Quiz #6-8 Nov 4, 9:45AM HW #1
3	Nov 9 (DC) - Linkage Disequilibrium - Genetic association studies	Nov 11 (DC) - Veterans' Day, no class	Nov 9 (in-class exercise 2, BT) LD link	Nov 9, 9:45AM Quiz #9-11 Nov 12, 9:45AM HW #2
4	Nov 16 (DC) - GWAS	Nov 18 (DC) - Population stratification w/DAG example.	Nov 18 (in-class exercise 3, AY) GWAS Catalog	Nov 16, 9:45AM Quiz #12
5	Nov 23 (DC/JH) - Power Calculations - Achievement of GWAS and Post-GWAS - Race/ancestry/ethnicity in genetic studies	Nov 25 (No class) - Thanksgiving Break	Nov 23 (in-class discussion 1) - Ethics consideration in genetic studies - NAS video, 10'	Nov 23, 9:45AM Review readings for class discussion on ethics Nov 23, 9:45AM HW #3 Nov 25, 9:45AM Group project draft
6	Nov 30 (Guest lecture) - Hugues Aschard, PhD Fine mapping; Polygenic risk score; Gene-environment interaction (GxE)	Dec 2 (Guest lecture/DC) - Hugues Aschard: GxE continued - Mendelian Randomization (MR) with power calculation	Dec 2 (in-class discussion 2) MR reading – Alcohol, BP, stroke paper	Dec 2, 9:45AM HW #4 Dec 2, Teaching staff returns comments on project drafts
7	Dec 7 (DC) - MR power (if needed) - Rare variants: discovery, population genetics, analysis - Rare variants: applications	Dec 9 (DC/Guest lecture) - Rare variants, applications/CNVs - Monica Wojcik, MD, MPH Genetic diagnosis in the clinic	Dec 7 (in-class exercise 4, BT) gnomAD Database	Dec 9, 9:45AM HW #5
8	Dec 14 (Guest lecturer) - Jordan Smoller, MD - Diversity in genetic studies: The All of Us resource - CNV/Optional time.	Dec 16 (Instructors/class) - Project Presentations	Dec 14 (In-class discussion 3) Possible discussion if time: WGS in biobanks; pan-ancestry PRS; EHR-based datasets vs. longitudinal cohorts	Dec 16, 9:45AM Group project final (proposal as *.docx or *.pdf; slides for presentation as *.ppt or *.pdf) Dec 16, 9:45AM HW #6