

Genetic Epidemiology
Spring Quarter 2022
EPI 517 / PHG 511

Course information web site: <https://canvas.uw.edu/courses/1547479>

Location: T-530 (Health Sciences Building)

Classes: M,W 11.30-12.50

Instructor: Sara Lindstroem (saralind@uw.edu)

TA: Austin Hammermeister Suger (hammea2@uw.edu)

Office Hours: Mondays and Wednesdays 1-2pm at HRC Conference Room 897 (8th floor of the Hans Rosling Center for Population Health)

Required Readings: 1 paper/lecture to be read before lecture. See Canvas for list of papers and access to pdf copies.

Other Recommended literature: Melissa A. Austin. Genetic Epidemiology: Methods and Applications.

Land Acknowledgement:

The University of Washington acknowledges the Coast Salish people of this land, the land which touches the shared waters of all tribes and bands within the Duwamish, Suquamish, Tulalip and Muckleshoot nations.

COVID-related expectations

Per UW policy, this class will be conducted in person. You should only register for this class if you can attend in person, or if you meet the criteria for an accommodation from Disability Resources for Students (DRS) or a special arrangement approved by the SPH Office of the Dean that allows you to take the course remotely.

- Please [contact UW Disability Resources for Students \(DRS\)](#) directly if you feel you may be eligible for an accommodation based on your status as an immunocompromised individual or based on other diagnosed physical or mental health conditions that might prevent you from being able to take classes in-person.
- If you are a student enrolled in a program in SPH, and you are either living with an individual who is immunocompromised, OR you are unable to obtain a visa to travel to the US, you may be eligible for a “special arrangement” that will allow you to take this course remotely. To further clarify, immunocompromised refers to individuals with no/critically weakened immune response to the vaccines. Immune compromised is not the same as underlying health concerns which may lead to a more severe response to COVID. Requests for special arrangements to take the class remotely should have been submitted to and approved by the Students and Academic Services team in the Office of the Dean before the beginning of the quarter.

All UW students are expected to complete their [vaccine attestation](#) before arriving on campus. [Per President Cauce’s message March 8](#), following changes to state and local health policies, masks will become optional inside most University facilities starting March 28, the first day of spring quarter. Masks will continue to be required in clinical and other health-care settings and on public transportation, including UW shuttles. We strongly recommend wearing masks indoors during the first two weeks of spring quarter. Please monitor yourself daily for symptoms and stay home if you are sick. It’s also strongly recommended to get tested after travel. Refer to the [UW Face Covering Policy](#) for the latest guidance and follow the campus-wide face-covering policy at all times. You are expected to follow state, local, and UW COVID-19 policies and recommendations. If you feel ill or exhibit possible COVID symptoms, you should not come to class. If you need to temporarily quarantine or isolate per CDC guidance and/or [campus policy](#), you are responsible for notifying your instructors as soon as possible by email. **If you receive a positive COVID-19 test result, you must report to campus Environmental Health & Safety (EH&S) by emailing covidehc@uw.edu or calling 206-616-3344.**

Please check your email daily BEFORE coming to class. If we need to conduct class remotely because the instructor or a guest speaker is complying with UW policies and unable to attend in person, we will send all registered students an email with a Zoom link for remote instruction.

Course Description:

This course focuses on the fundamental research methods and applications in the dynamic field of genetic epidemiology. By the end of the course, students will be familiar with key genetic concepts, describe current genomic technologies that are used in genetic epidemiology, have an understanding of various study designs in genetic epidemiology and be able to identify relevant software and web-based data resources.

Topics Covered:

- Approaches for assessing genetic influences on complex traits
- Advances in genomics technology
- Population-based and family-based study designs
- Candidate gene and genome-wide association studies of both common and rare genetic variants
- Gene-environment interaction studies
- Mendelian Randomization studies
- Risk Prediction

Learning Objectives:

1. Explain key genetic concepts
2. Describe current genomic technologies that are used in genetic epidemiology
3. Conduct analysis of genetic data using PLINK
4. Describe and compare research study designs used in genetic epidemiology, including family-based studies, genetic association studies, and studies of gene-environment interactions
5. Discuss recent literature in genetic epidemiology

Course Format:

Lectures, computer exercises, home assignments, and readings

Grading structure:

Student Presentations	30%
Recaps of previous lectures, journal club and active participation	
Home assignments	40%
Take-home exam	30%

Homework assignments

There will be four home assignments throughout the course (due dates 4/14, 4/28, 5/12, 5/26). These will include theoretical questions as well as R exercises to give you hands-on experience of analyzing genetic data. We will have an intro lecture to R the second week of the course. Home assignments should be submitted via the CANVAS site by midnight of the due date. You will have one week (seven days) to complete the home assignments.

Feedback to the Instructors

We welcome any comments, suggestions, criticisms, or compliments you care to offer about the course as we go along. Students with concerns about a seminar speaker, instructor, or TA should discuss it with those individuals. If the student is not satisfied with the response, they may contact the Chair of the Department of Epidemiology. If concerns are not satisfactorily resolved, the student may also contact the Graduate School. Online course evaluations will be available in June.

Schedule

	Lecture	Day	Date	Instructor	Topic
Week 1	Lecture 1	Mon	28-Mar	Sara	Overview of Genetic Epi
	Lecture 2	Wed	30-Mar	Sara	Human Genetic Variation
Week 2	Lecture 3	Mon	4-Apr	Austin	Introduction to PLINK
	Lecture 4	Wed	6-Apr	Sara	Linkage disequilibrium and HWE
Week 3	Lecture 5	Mon	11-Apr	Sara	Genotyping, Imputation, Sequencing
	Lecture 6	Wed	13-Apr	Sara	Population differences, admixture mapping
Week 4	Lecture 7	Mon	18-Apr	Sara	Analysis of genetic association studies
	Lecture 8	Wed	20-Apr	Sara	Genome-wide association studies (GWAS)
Week 5	Lecture 9	Mon	25-Apr	Austin	Rare variant analyses
	Lecture 10	Wed	27-Apr	Sara	Family-based tests
Week 6	Lecture 11	Mon	2-May	Sara	Gene-Environment interactions 1
	Lecture 12	Wed	4-May	Sara	Gene-Environment interactions 2
Week 7	Lecture 13	Mon	9-May	Sara	Ethics in genetic epidemiology
	Lecture 14	Wed	11-May	Stephanie	Functional annotations
Week 8	Lecture 15	Mon	16-May	Jessica/Kristina	The Microbiome and DNA Methylation
	Lecture 16	Wed	18-May	Jeroen	Genetics and Epidemiology of Colorectal Cancer Consortium (GECCO)
Week 9	Lecture 17	Mon	23-May	Sara	Mendelian Randomization
	Lecture 18	Wed	25-May	Sara	Risk prediction
Week 10		Mon	30-May	Memorial Day	N/A
	Lecture 19	Wed	1-Jun	Sara	Review

Required Readings

	Day	Date	Required Reading
Week 1	Mon	28-Mar	
	Wed	30-Mar	A brief history of human disease genetics.
Week 2	Mon	4-Apr	R intro documentation
	Wed	6-Apr	Patterns of linkage disequilibrium in the human genome.
Week 3	Mon	11-Apr	Genotype Imputation from Large Reference Panels
	Wed	13-Apr	Race and Genetic Ancestry in Medicine - A Time for Reckoning with Racism
Week 4	Mon	18-Apr	A tutorial on statistical methods for population association studies
	Wed	20-Apr	Genomewide association studies for complex traits: consensus, uncertainty and challenges.
Week 5	Mon	25-Apr	Rare-variant association analysis: study designs and statistical tests.
	Wed	27-Apr	Family-based designs in the age of large-scale gene-association studies.
Week 6	Mon	2-May	Gene-environment interactions for complex traits: definitions, methodological requirements and challenges.
	Wed	4-May	Update on the State of the Science for Analytical Methods for Gene-Environment Interactions
Week 7	Mon	9-May	Paper 1: Genomic Justice for Native Americans: Impact of the Havasupai Case on Genetic Research Paper 2: Large-scale GWAS reveals insights into the genetic architecture of same-sex sexual behavior. Paper 3: The Ethics of Big Data in Genomics: The Instructive Icelandic Saga of the Incidentalome
	Wed	11-May	N/A
Week 8	Mon	16-May	N/A
	Wed	18-May	N/A
Week 9	Mon	23-May	Recent Developments in Mendelian Randomization Studies.
	Wed	25-May	Clinical use of current polygenic risk scores may exacerbate health disparities
Week 10	Mon	30-May	N/A
	Wed	1-Jun	N/A

Academic Integrity

Students at the UW are expected to maintain the highest standards of academic conduct, professional honesty, and personal integrity.

The UW School of Public Health (SPH) is committed to upholding standards of academic integrity consistent with the academic and professional communities of which it is a part. Plagiarism, cheating, and other misconduct are serious violations of the University of Washington [Student Conduct Code \(WAC 478-121\)](#). We expect you to know and follow the university's policies on cheating and plagiarism, and the [SPH Academic Integrity Policy](#). Any suspected cases of academic misconduct will be handled according to University of Washington regulations. For more information, see the University of Washington [Community Standards and Student Conduct](#).

Access and Accommodations

Your experience in this class is important to me. It is the policy and practice of the University of Washington to create inclusive and accessible learning environments consistent with federal and state law. If you have already

established accommodations with Disability Resources for Students (DRS), please activate your accommodations via myDRS so we can discuss how they will be implemented in this course.

If you have not yet established services through DRS but have a temporary health condition or permanent disability that requires accommodations (conditions include but not limited to; mental health, attention-related, learning, vision, hearing, physical or health impacts), contact DRS directly to set up an Access Plan.

DRS facilitates the interactive process that establishes reasonable accommodations. Contact DRS at disability.uw.edu.

Religious Accommodations

Washington state law requires that UW develop a policy for accommodation of student absences or significant hardship due to reasons of faith or conscience, or for organized religious activities. The UW's policy, including more information about how to request an accommodation, is available at [Religious Accommodations Policy \(https://registrar.washington.edu/staffandfaculty/religious-accommodations-policy/\)](https://registrar.washington.edu/staffandfaculty/religious-accommodations-policy/). Accommodations must be requested within the first two weeks of this course using the [Religious Accommodations Request form \(https://registrar.washington.edu/students/religious-accommodations-request/\)](https://registrar.washington.edu/students/religious-accommodations-request/).

Inclusion and Diversity:

Diverse backgrounds, embodiments and experiences are essential to the critical thinking endeavor at the heart of University education. In SPH, we are expected:

1. To respect individual differences, which may include, but are not limited to, age, cultural background, disability, ethnicity, family status, gender, immigration status, national origin, race, religion, sex, sexual orientation, socioeconomic status and veteran status.
2. To engage respectfully in the discussion of diverse worldviews and ideologies embedded in course readings, presentations and artifacts, including those course materials that are at odds with personal beliefs and values.
3. To encourage students with concerns about classroom climate to talk to their instructor, adviser, a member of the departmental or SPH EDI Committee, the Assistant Dean for EDI, or the program's director.

Classroom Climate:

We are co-creators of our learning environment. It is our collective responsibility to develop a supportive learning environment for everyone. Listening with respect and an open mind, striving to understand others' views, and articulating your own point of view will help foster the creation of this environment. We engage our differences with the intent to build community, not to put down the other and distance our self from the other. Being mindful to not monopolize discussion and/or interrupt others will also help foster a dialogic environment.

The following guidelines can add to the richness of our discussion:

- We assume that persons are always doing the best that they can, including the persons in this learning environment.
- We acknowledge that systematic oppression exists based on privileged positions and specific to race, gender, class, religion, sexual orientation, and other social variables and identities.
- We posit that assigning blame to persons in socially marginal positions is counter-productive to our practice. We can learn much about the dominant culture by looking at how it constructs the lives of those on its social margins.
- While we may question or take issue with another class member's ideology, we will not demean, devalue, or attempt to humiliate another person based on her/his experiences, value system, or construction of meaning.

- We have a professional obligation to actively challenge myths and stereotypes about our own groups and other groups so we can break down the walls that prohibit group cooperation and growth.
[Adapted from Lynn Weber Cannon (1990). Fostering positive race, class and gender dynamics in the classroom. *Women Studies Quarterly*, 1 & 2, 126-134.]

We are a learning community. As such, we are expected to engage with difference. Part of functioning as a learning community is to engage in dialogue in respectful ways that supports learning for all of us and that holds us accountable to each other. Our learning community asks us to trust and take risks in being vulnerable.

Here are some guidelines that we try to use in our learning process:

- LISTEN WELL and be present to each member of our group and class.
- Assume that I might miss things others see and see things others miss.
- Raise my views in such a way that I encourage others to raise theirs.
- Inquire into others' views while inviting them to inquire into mine.
- Extend the same listening to others I would wish them to extend to me.
- Surface my feelings in such a way that I make it easier for others to surface theirs.
- Regard my views as a perspective onto the world, not the world itself.
- Beware of either-or thinking.
- Beware of my assumptions of others and their motivations.
- Test my assumptions about how and why people say or do things.

Be authentic in my engagement with all members of our class.

Pronouns:

We share our pronouns because we strive to cultivate an inclusive environment where people of all genders feel safe and respected. We cannot assume we know someone's gender just by looking at them. So we invite everyone to share their pronouns.

Bias Concerns:

The Office of the Dean has a [student concern policy](#), a faculty concern policy and standard HR procedures for staff concerns. Our 2018 climate survey states that most people in SPH do not report bias incidents because they do not know where to go. Students are encouraged to report any incidents of bias to someone they feel comfortable with, including instructors, advisers or department staff. They can email dcinfo@uw.edu for immediate follow up. Bias concerns can be anonymously and confidentially reported at <https://sph.washington.edu/about/diversity/bias-concerns>. Data is collected by the Assistant Dean for EDI and the Director of Program Operations for Student and Academic Services and tracked for resolution and areas are identified for further training.

Sexual Harassment:

Sexual harassment is a form of harassment based on the recipient's sex that is characterized by:

1. Unwelcome sexual advances, requests for sexual favors, or other verbal or physical conduct of a sexual nature by a person who has authority over the recipient when:
 - Submission to such conduct is an implicit or explicit condition of the individual's employment, academic status, or ability to use University facilities and services, or
 - Submission to or rejection of the conduct affects tangible aspects of the individual's employment, academic status, or use of University facilities.

2. Unwelcome and unsolicited language or conduct that creates an intimidating, hostile, or offensive working or learning environment, or has the purpose or effect of unreasonably interfering with an individual's academic or work performance.

If you believe that you are being harassed, or have observed harassment, you can report it to SPH using the [bias concerns link](#). The University also has designated offices to help you: [SafeCampus](#); [Office of the Ombud](#); [Title IX Investigation Office](#); and [University Complaint Investigation and Resolution Office](#).

Campus safety:

Call SafeCampus at 206-685-7233 anytime – no matter where you work or study – to anonymously discuss safety and well-being concerns for yourself or others. SafeCampus's team of caring professionals will provide individualized support, while discussing short- and long-term solutions and connecting you with additional resources when requested.

Mental Health Resources:

Your mental and physical health are at the forefront of our minds. Please do not hesitate to tell us about any challenges you may be facing. UW students have access to several resources on/off campus. These include individual counseling, group therapy, workshops and crisis services among other things. For a complete list of resources, please go to <https://wellbeing.uw.edu/topic/mental-health/>. Students currently enrolled in degree-seeking programs at the UW Seattle campus are generally eligible for services at the Counseling Center (<https://www.washington.edu/counseling>), which offers individual and group counseling free of charge.