SCHOOL OF PUBLIC HEALTH
EPIDEMIOLOGY
UNIVERSITY of WASHINGTON

WELCOME

SISG 2023: Module 11 Genetic Epidemiology

Land Acknowledgement

We acknowledge that University of Washington stands on the unceded land of the Coast Salish peoples, land which touches the shared waters of all tribes and bands within the Duwamish, Suquamish, Tulalip and Muckleshoot nations

Burcu Darst

Assistant professor
Public Health Sciences Division, Fred Hutchinson Cancer Center

Pronouns: she, her, hers

I am a genetic epidemiologist research is focused on identifying and understanding genetic and multi-omic risk factors of prostate cancer and other complex traits across diverse populations. In particular, my research uses rare variants captured with whole exome sequencing, genome-wide association studies, polygenic risk scores, and metabolomics to investigate disease risk.

Sara Lindström

Associate professor
Department of Epidemiology, UW
Public Health Sciences Division, Fred Hutchinson Cancer Center

Pronouns: she/her/hers

I am a genetic epidemiologist with a special interest in understanding the genetic contribution to common complex diseases, with a primary emphasis on cancer. By leveraging large population-based studies, I investigate how our genetics and environment affect disease risk.

I also teach PHG511/EPI517 “Genetic Epidemiology” as part of the UW Public Health Genetics program and the UW Department of Epidemiology.

Schedule

Wednesday July 19

1.30-2.00 Session 1. Introductions/Logistics. **Sara and Burcu**
2.00-3.00 Session 2. Introduction to Genetic Epidemiology. **Sara.**
3.00-3.30 Break
3.30-4.30 Session 3. Types of human genetic variation. **Burcu**
4.30-5.00 Q&A

Thursday July 21

9.00-10.00 Session 4. LD and HWE. **Sara.**
10.00-10.30 Break
10.30-12.00 Session 5. Assessing genetic variation, imputation, principal component analysis. **Burcu.**
12.00-1.30 Lunch
1.30-2.15 Session 6. Analysis of association studies. **Sara.**
2.15-3.00 Session 7. Genome-wide association studies. **Burcu.**
3.00-3.30 Break
3.30-4.30 Session 8. Rare variants. **Sara.**
4.30-5.00 Q&A

Friday July 21

9.00-10.00 Session 9. Risk prediction. **Burcu.**
10.00-10.30 Break
10.30-12.00 Session 10. Ethics in Genetic Epidemiology. **Sara and Burcu.**
12.00-1.30 Lunch
1.30-3.00 Session 11. Gene-Environment interactions. **Sara.**
3.00-3.30 Break
3.30-4.30 Session 12. Mendelian Randomization. **Burcu.**
4.30-5.00 Q&A

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SISG website

- We will use the SISG website as the main source of information. Here you can find slide decks from lectures, recorded lectures, recommended readings, exercises etc.

- https://si.biostat.washington.edu/institutes/sisg/SM2311

Q&A

- We will have Q&A at the end of each day 4.30-5pm.

- These will provide an opportunity to ask the instructors any questions you might have about course content or genetic epidemiology in general (they will also give us some buffer time in case we didn't have time to cover everything during class)

- If you want to request a 1-on-1 appointment with one of the instructors during Q&A, please let us know

Recommended Readings

> To get as much as possible out of this Module, we have compiled a set of papers that we recommend you read for more in-depth information.

Topic	Reading
Genetic Epi overview	A brief history of human disease genetics.
Types of human genetic variation	Sequencing of 53,831 diverse genomes from the NHLBI TOPMed Program
Hardy-Weinberg Equilibrium and Linkage Disequilibrium	Patterns of linkage disequilibrium in the human genome.
Assessing genetic variation, imputation, principal component analysis	Race and Genetic Ancestry in Medicine – A Time for Reckoning with Racism
Analysis of association studies	Genome-wide association studies
Genome-wide association studies	15 years of GWAS discovery: Realizing the promise
Rare variants	Rare-variant association analysis: study designs and statistical tests.
Risk prediction	From Basic Science to Clinical Application of Polygenic Risk Scores: A Primer
Gene-Environment interactions	Gene-environment interactions for complex traits: definitions, methodological requirements and challenges.
Mendelian Randomization	Statistical methods for Mendelian randomization in genome-wide association studies: A review.

Slack

- We have set up a Slack channel that allows for one-to-one direct messaging between participants and instructors, as well as the option to set up a group channel where multiple members can chat, share files etc.

- Information about downloading and using slack (either on your computer and/or phone) can be found at https://slack.com (Links to an external site.).

- You should have received an email invitation to join the mod11_genetic_epidemiology_2023 slack channel

Session 10: Ethics in Genetic Epidemiology

- You will be divided into 3 groups
- We will spend 15 mins on each paper: 10 mins in group and 5 minutes as a large group.
  - For paper 1, group 1 will report on their discussion to the large group
  - For paper 2, group 2 will report on their discussion to the large group
  - For paper 3, group 3 will report on their discussion to the large group
- Everyone will read and discuss all 3 papers
- For each paper, 3 questions will be posted to help facilitate discussions

Session 10: Ethics in Genetic Epidemiology

- **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* (only read Chapters1-4; pages 351-365)

Breakout Activity

We will utilize break-out sessions with groups of ~4 throughout the module to enhance the opportunity to conduct practical assignments and discuss relevant topics with your peers.

1. Each person, please introduce yourself to the other members in your group:
   - Your name and pronouns. Your position (student, researcher) and affiliation (what University or institute).
   - What are your strengths in your training so far? (i.e., is your background in genetics, biostatistics, law?)
   - What prompted you to take this course? What are you hoping to learn?

2. Once everyone is introduced, discuss in your group:
   - Why do we study the role of genetics in human disease?