

WELCOME

SISG 2021:  
Module 10  
Genetic Epidemiology

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

*-University of Washington land acknowledgement*

# Housekeeping

- Introductions of instructors
- Schedule
- Layout of lectures – Zoom poll, breakout rooms
- Slack channel
- Q&A at the end of Thursday/Friday
- Zoom polls (What is genetic epi, what is your current role (student, postdoc, faculty, scientist at research institute, industry), which time zone are you in)



## Alison (Alie) Fohner

---

Pronouns: she, her, hers

Dr. Fohner is a genetic epidemiologist applying innovative big data methods to extensive electronic health record, proteomic, and genomic data to improve precision medicine. She approaches genetic epidemiology through an ELSI (ethical, legal, social implications) lens, and aims to promote equity and justice in the benefit of and access to genetic information in health settings. She teaches in the Institute of Public Health Genetics, an interdisciplinary program in the School of Public Health that explores the application of genomics in public health.



Sara Lindström

Associate professor

Department of Epidemiology, UW

Public Health Sciences Division, Fred Hutchinson Cancer  
Research 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 and linked traits. 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.



Hanley Kingston (TA)

PhD Candidate

Institute of Public Health Genetics, UW

Pronouns: she / they

I am a second/third-year PhD student at the school of public health genetics, UW. My research involves investigating confounding by population structure in genome-wide association studies and conducting phylogenetic analyses of HIV and hepatitis C. I am excited to be your TA for this class!

Day	Time	Lead	Topics	Details
Wed	11:45-12.30	Alie/Sara	Class Intro	Intro to class/agenda and topics Technology/logistics Breakout room introductions
<b>BREAK</b>				
	12.45-1.30	Sara	Overview of Genetic Epi	Intro to Epidemiology and Genetic Epidemiology
<b>BREAK</b>				
	1:45-2:30	Alie	Human genetic variation	Types of genetic variation

**THURSDAY****8:30 – 9:15**

Alie

HWE/LD

Hardy Weinberg Equilibrium, and Linkage Disequilibrium

**BREAK****9:30-10:15**

Alie

Population Structure

Ancestry and Principal Component Analysis

**BREAK****10:30-11:15**

Sara

Study Designs

Types of genetic epidemiology studies, imputation

**LUNCH BREAK****11:45 – 12:30**

Alie

Association studies

Conducting association studies and calculating odds ratios

**BREAK****12:45-1:30**

Sara

GWAS

Genome wide association studies

**BREAK****1:45 – 2:30**

Sara/Alie

Office Hours

Stop by to ask questions from the day, or schedule time to discuss your own project.



**FRIDAY****8:30 – 9:15**

Alie

Bioethics/  
ImplementationBioethical principles in genetic epidemiology,  
deciding whether to implement genetic testing**BREAK****9:30-10:15**

Sara

Rare variation

Strategies to analyze rare genetic variants

**BREAK****10:30-11:15**

Sara

GxE interactions

Gene x Environment interactions analyses

**LUNCH BREAK****11:45 – 12:30**

Alie

Risk prediction

Polygenic risk scores and population screening

**BREAK****12:45-1:30**

Sara

Mendelian  
Randomization

Mendelian Randomization studies

**BREAK****1:45 – 2:30**

Sara/Alie

Office Hours

Stop by to ask questions from the day, or  
schedule time to discuss your own project.

# 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/suminst/SISG2021/modules/SM2110>

# Lecture recordings

- We will be recording the sessions. Only people with paid registration will have access to the recording links.
- For requests (i.e., closed captions, transcriptions, etc.), contact Hanley via Slack.

# Office Hours

- We will have Office Hours Thursday and Friday 1.45-2.30pm PT.
- These will provide an opportunity to ask the instructors any questions you might have. These sessions are optional. You can stop by to ask general questions or just listen in on discussion.
- If you want to request a 1-on-1 appointment with one of the instructors during Office Hours, there will be 10-mins slots that you can sign up for via Slack by direct messaging Hanley, our TA

# Recommended Readings

- To get as much as possible out of this Module, we have compiled a set of papers that we recommend that you read for more in-depth information and to see some of our favorite examples of the methods we will discuss.

Session	Topic	Reading	PMID
2	Genetic Epi overview	A brief history of human disease genetics.	31915397
3	Human Genetic Variation	Sequencing of 53,831 diverse genomes from the NHLBI TOPMed Program	33568819
4	HWE/LD	Patterns of linkage disequilibrium in the human genome.	11967554
5	Ancestry and Principal Component Analysis	Race and Genetic Ancestry in Medicine – A Time for Reckoning with Racism	33406325
6	Types of genetic epidemiology studies, imputation	Genotype Imputation from Large Reference Panels	29799802
7	Conducting association studies and calculating odds ratios	A tutorial on statistical methods for population association studies;	16983374
8	Genome wide association studies	Genome-wide association studies for complex traits: consensus, uncertainty and challenges.	18398418
9	Bioethical principles in genetic epidemiology	<a href="https://depts.washington.edu/bhdept/ethics-medicine/bioethics-topics/articles/principles-bioethics">https://depts.washington.edu/bhdept/ethics-medicine/bioethics-topics/articles/principles-bioethics</a>	
10	Strategies to analyze rare genetic variants	Rare-variant association analysis: study designs and statistical tests.	24995866
11	Gene x Environment interactions analyses	Gene-environment interactions for complex traits: definitions, methodological requirements and challenges.	18523454
12	Polygenic risk scores and population screening	Clinical use of current polygenic risk scores may exacerbate health disparities	30926966
13	Mendelian randomization	Recent Developments in Mendelian Randomization Studies.	29226067

# Break-out sessions

- We will utilize Zoom break-out sessions with groups of 4-5 throughout the module to enhance the opportunity to conduct practical assignments and discuss relevant topics with your peers.
- We will assign you a break-out room at the beginning of each day and will update the roster every day to give you a chance to meet different peers for networking.
- If you have questions, send a note in slack identifying your group number. Instructors will rotate through the groups.
- One member of each group, please post group results in slack.

We will also use Zoom polling to complete smaller activities.

- Practice zoom poll

# 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 will receive an email invitation to join the *md10\_genetic\_epidemiology\_2021* Slack Channel.



# Zoom chat

- Please use the chat function to ask questions for quick answers. Hanley will be monitoring the chat and slack channel throughout the module.

# Breakout Activity

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?